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Caring for a Child with Congenital Adrenal Hyperplasia: Health-related Quality of Life and Needs of the Parents

Dissertation

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1. Introduction

Congenital adrenal hyperplasia signifies a rare autosomal recessive passed endocrine disorder. Its most common form is based on a deficiency of the 21-hydroxylase enzyme [21-OH], accounting for approximately 95% of the cases. Classic congenital adrenal hyperplasia with 21-OH deficiency [in the following simplified labelled as CAH] is the result of deletions or deleterious mutations in the active gene CYP21A2, which cause varying degrees of impairment of 21-OH activity. The worldwide incidence lies between 1:14000 to 1:18000 births. Affected female new-borns can show ambiguous external genitalia at birth and in both sexes with the salt-wasting form, hazardous adrenal crises developed postnatally (Stokowski, 2009, White and Bachega, 2012, Speiser et al., 2018). Mortality associated with CAH in children is reported to be in the range between 3% and 13% (Swerdlow et al., 1998, Lim et al., 1995). Not only because of its life-threatening character it is essential to discover CAH early and thus reduce grave consequences. With admission to new-born screening in many countries worldwide, CAH can now be detected using dried blood on filter paper (Dörr et al., 2015).

The diagnosis of a rare chronic disease such as CAH constitutes a profound change in parental thinking. The expectant mothers and fathers must deal not only with the new task of being a parent but also with the loss of their idea of giving birth to ‘the perfect child’. A lot of studies verify an increasing psychosocial burden in the entire family after a new-born gets the diagnosis of a chronic disease (Satterwhite, 1978, Grootenhuis and Bronner, 2009, Cousino and Hazen, 2013, Sundus et al., 2013). However, less is known about the psychosocial situation of parents giving birth to a child with CAH. A few studies point out an increasing strain in these parents, especially in the time shortly after the final diagnosis of their new-born is given (Stokowski, 2009, Barg et al., 2010, Fleming et al., 2017b, Witt et al., 2018, Waldthausen, 2007). A high level of parental distress might endanger the development of a stable parent-child-relationship, which is though important for a healthy upbringing of the child. Furthermore, parental difficulties in dealing with their child’s disease may lead to problems in the development of the child’s coping skills later on (Grossmann and Grossmann, 2007, Grootenhuis and Bronner, 2009, Grossmann, 2010, Wiegand-Grefe et al., 2016, Gentile and Fusco, 2017).

To improve the psychosocial situation of the parents caring for a child diagnosed with CAH and thus build a reasonable basis for a healthy upbringing, it is crucial to

understand the needs of affected families. Gathering the dimensions of the health-related quality of life [HrQoL], the coping patterns and the specific needs of affected parents allow clinicians and researchers to gain insight into the life of these families. With this knowledge, it will be possible to develop demand-responsive, purposeful interventions, especially for the time facing the diagnosis. Therefore, this study aims to examine the following questions:

1) How does the HrQoL of parents caring for a child diagnosed with CAH differ from reference populations?
2) Which coping patterns and special needs do these parents have, and is there a correlation between successful coping or the intensity of needs and the established HrQoL?

2. Theoretical Background

To understand the situation of parents caring for a child diagnosed with CAH, it is helpful to know about the circumstances of becoming a parent in general and the difficulties of caring for a chronically ill child as well as main facts about the disease pattern of CAH. For proper research on the HrQoL, coping strategies and needs of parents caring for CAH diagnosed children, it is additionally essential to know the current state of this topic.

2.1 Transition to Parenthood in a Psychosocial Context

The announcement of a pregnancy is mostly a fortunate event. But dealing with it and facing the new tasks of upcoming parenthood can be very troublesome for the expectant parents. Gentile and Fusco (2017, p. 325) state, that the ”transition to parenthood represents an important life event which increases vulnerability to psychological disorders“.

Already in times of pregnancy, parents can experience psychological strain like anxiety, distress, and depression (Morse et al., 2000, Parfitt and Ayers, 2014). This does not change severely in the first months after giving birth. Especially first-time parents are at high risk for developing postpartum depression and parenting distress (Epifanio et al., 2015). Epifanio et al. (2015, p. 38) describe the first month post-partum as ”[…] a critical phase of parents life […]“ and ”[…] a developmental crisis

characterized by anxiety, stress and mood alterations that could have important repercussions on the child psycho-physical development […]".

Beside this psychological challenge, the new parents must deal with further difficulties such as magnified financial problems and sleep disturbances. Caring for a child can cause financial strain for parents through additional expenses, e.g. for the children's food, clothing, and medical care. The sleep disturbances appear according to Nelson et al. (2014), especially in parents with younger children.

Another substantial change concerns the relationship between the expectant couple. Cox (1985, p. 402) declares that "[…] parenthood causes significant change and reorganization in the couple system". The new mothers and fathers have to deal with reduced satisfaction and quality of their relationship in general (Delicate et al., 2018) as well as new couple's role arrangements, increasing numbers of conflicts (Cowan et al., 1985), and a deterioration of their sexual relationship (Parfitt and Ayers, 2014).

Summarizing all, expectant parents must undergo increasing psychological pressure, difficulties in external life circumstances like finances, and a changing couple's relationship during the time of pregnancy and the first months postpartum.

2.2 Caring for a Chronically Ill Child

Besides the mental load of arranging a new family life system, it is an extraordinary challenge to give birth to a new-born with a chronic health condition (Cousino and Hazen, 2013). The diagnosis of a chronic disease of a new-born is often a shocking occurrence associated with a high level of emotional distress, especially for the parents (Fonseca et al., 2013). A lot of new, unexpected additional responsibilities are approaching the new mothers and fathers. These could be, e.g. additional care tasks, including medical care and drug application, a stricter financial management, thoughtful planning of daily activities, and much more (Grootenhuis and Bronner, 2009). Since the distinctive characteristic of a chronic illness is its incurability, affected children require lifelong care. Therefore, these new responsibilities for the parents often last a lifetime and the new mothers and fathers definitely have to let go of their dream of the 'perfect new-born'.

If the child is diagnosed with a rare disease, parents are faced with even more additional strains (Dellve et al., 2006). According to the World Health Organization [WHO] (2013, p. 1) a rare disease is defined by a prevalence of less than 5 per

10000 inhabitants and is accompanied by "[…] fundamentally different challenges from those of more common diseases […]". Parents must overcome difficulties in obtaining a correct diagnosis and in finding information about the rare disease. Moreover, there is a shortage of specialized health care services for rare diseases, which result in further challenges (Jaffe et al., 2010).

In summary, parents with a child diagnosed with a rare chronic health condition seem to have a much higher level of emotional pressure than parents caring for a healthy new-born. Next to the primary responsibilities of new parents, they have to deal with additional disease-specific tasks to enable a healthy development for their child. Some studies examining the situation of families with a paediatric chronic disease point out, that the parents in those families have to adjust to restrictions in their social life, job perspective, and psychosocial well-being as their everyday life is decisively determined by the child's care responsibilities (Grootenhuis and Bronner, 2009, Nehring et al., 2015). According to Nehring et al. (2015) the parental adaptation to the new situation of having a chronically ill child often happens without a lot of support for coping with the child's diagnosis. Supplies of psychosocial support are rarely offered, which makes it additionally hard to assimilate the new family situation.

2.3 Congenital Adrenal Hyperplasia with 21-Hydroxylase Deficiency [CAH]

To fully retrace the situation of families concerned by CAH, it is crucial to understand this disease pattern within its complexity. CAH is an endocrinal chronic disease arising from dysfunction of adrenal steroidogenesis. It corresponds to a group of inherited autosomal recessive disorders that result from a deficiency in one or several of the enzymes of cortisol biosynthesis. With a worldwide incidence of 1:14000 to 1:18000 births, it is counted among the 5000 to 8000 existing rare diseases (WHO, 2013, Speiser et al., 2018).

The most common form of congenital adrenal hyperplasia is a deficiency of the 21-OH, accounting for approximately 95% of all cases (Speiser et al., 2018). The 21-OH deficiency is the result of deletions or deleterious mutations in the active gene CYP21A2, which cause varying degrees of impairment of 21-OH activity. Depending on the remaining enzyme activity, the clinical phenotype of CAH occurs as 'classic CAH' for the severe form, and 'non-classic CAH' for the mild form. Classic CAH is further classified into two main phenotypes: the three times more frequent appearing

(1) CAH with salt-wasting and the (2) simple-virilising type without salt-losing syndrome (Dörr and Schulze, 1998, White and Bachega, 2012, Stokowski, 2009). Both types have ambiguous genitalia in new-born girls in common. Patients affected by the salt-wasting type (1) exhibit further life-threatening adrenal crises, appearing for the first time usually between the second and third week after birth. Male new-borns with simple-virilising CAH and girls with CAH not diagnosed at birth show pseudo-precocious puberty from infancy on (Dörr and Schulze, 1998, Stokowski, 2009). In untreated or not adequately treated CAH children, there is an acceleration of bone maturation and premature closure of the epiphyses, which results in extended growth in childhood and stunted growth in adult patients (Dörr and Schulze, 1998). According to its chronic and complex character, CAH is a lifelong disease impacting body weight, blood pressure, metabolism, bone mineral density, fertility, sexuality, and quality of life (Finkielstain et al., 2012, Malouf et al., 2010).

With the new-born screening there exists an effective method to detect CAH early, and thus provide timely and effective therapy and support mechanisms for affected families. The screening procedure is based on determination of 17-hydroxyprogesteron - a precursor of cortisol and aldosterone catalysed by 21-OH - in dried blood on filter paper taken on the third day of the new-born's life (Stokowski, 2009, Dörr et al., 2015). This way, children with the severe salt-wasting type are detected early and adrenal crises or death can be counteracted. Further, the new-born screening enables a reduction of the period of incorrect gender assignment in females with the simple-virilising type (Dörr et al., 2015).

Treatment of choice is a lifelong, daily substitution with glucocorticoids, e.g. hydrocortisone, and in all salt-wasting CAH forms additionally mineralocorticoids like fludrocortisone. The therapy management of children with CAH constitutes a challenge for parents and caregivers. They have to provide and monitor the daily hydrocortisone and fludrocortisone dosages, supplement maintenance dosages with oral 'stress dosing' during times of illness and in case of emergency apply intramuscular injection of glucocorticoids when a child is unable to tolerate oral medications and/or if signs of adrenal crisis are present (Merke and Bornstein, 2005). Since the needs for stress dosing are frequent and unpredictable, the treatment decisions are complex and cause high levels of distress in parents. Former studies suggest that mothers and fathers of CAH affected children are often overstrained with this situation and experience overtaxing and powerlessness

(Fleming et al., 2017a, Lundberg et al., 2017). Therefore, psychological support for the whole family is considered desirable from infanthood on to accompany the patient and the patient's environment in all future problems and difficulties and thus support the medical practitioner in achieving a comprehensive care (Dörr and Schulze, 1998, Witt et al., 2018).

2.4 Parents of Children with CAH - Psychosocial Aspects

Caring for a child with CAH constitutes an extraordinary challenge for delivering parents and caregivers. It is thus essential to take a closer look at the HrQoL, coping strategies, and parental needs to analyse their situation and estimate their strengths and difficulties in attending their child. Knowing this, demand-responsive interventions for parents caring for a child with CAH can be generated.

2.4.1 Health-related Quality of Life [HrQoL]

The concept of HrQoL had its advent in the medical field around 50 years ago in the 1970s (Bullinger, 2014). Based on the WHO's definition of health as "[…] a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity" (WHO, 2006, p. 1) researches then began to expand around the definition, measuring instruments and the practical importance of HrQoL. As Bullinger and Quitmann (2014) state, there is an ongoing paradigm shift in the medical field from measuring the clinical outcome of a patient based on clinical values towards patient-reported outcomes. HrQoL is a multidimensional patient-reported outcome, which assesses the subjective experienced well-being and health of a patient. In medical research, the most elaborated core dimensions of HrQoL are physical, social, functional, and mental health at which the latter can be seen as a generic term for emotional and cognitive health (Schnürch, 1995, Bullinger, 2014, Bullinger and Quitmann, 2014). Instruments for accessing HrQoL can be divided into generic, chronic-generic, and condition-specific approaches. Within this study, chronic-generic questionnaires are being used since they constitute an appropriate approach for accessing HrQoL of parents of chronically ill children as the results are easily comparable to other cohorts and sensitive to disease-related changes in HrQoL (Silva et al., 2013).

The current state of research on the subject 'HrQoL of parents of children diagnosed with CAH' is very sparsely as seen in the table below *(Table 1)*. There are no

published studies treating this specific topic. However, one unpublished study can be found. In 2007, Waldthausen wrote a doctoral thesis about the psychosocial situation of families with a child diagnosed with CAH, in which the parental HrQoL was examined as well. Contrary to previous expectations, Waldthausen discovered merely a slight reduction in the HrQoL of the 41 examined mothers and fathers and suggested that the parents in this study already exceeded the point of maximum strain since the mean age of their children was four years (Waldthausen, 2007). This consideration is confirmed by further studies (Suorsa et al., 2015, Ellens et al., 2017, Wolfe-Christensen et al., 2017, Witt et al., 2018).

However, there are some studies taking a closer look at the early psychosocial functioning of parents of children with a disorder of sex development [DSD] in general (Suorsa et al., 2015, Ellens et al., 2017, Wolfe-Christensen et al., 2017). The results of the study of Suorsa et al. (2015) show a significant reduction of the parental HrQoL concerning especially the core dimension of mental health. The children of the surveyed parents were on average ten months old, 44.8% were diagnosed with CAH, 24.1% with unclassified DSD and 31% with mixed gonadal dysgenesis and other DSD (Suorsa et al., 2015). After one year, a significant improvement of the parental HrQoL could be witnessed (Ellens et al., 2017). These findings relate to the results of the pilot study of Witt et al. (2018), surveying parents of children with CAH, which reveal a high level of perceived disease-related stress in respective parents, especially at the time of diagnosis. Barg et al. (2010) identify a specific burden in parents of children born with CAH right after receiving the diagnosis, as well. Many parents reported to have anxiety symptoms, feelings of helplessness, and worries about the future at that specific point in time (Barg et al., 2010).

Additionally, according to Suorsa et al. (2015) and Pasterski et al. (2014), parents of children with a DSD with ambiguous genitalia often suffer from post-traumatic stress symptoms after the disclosure of the diagnosis of their new-born. In the study of Pasterski et al. (2014), 31% of the mothers and 18% of the fathers were affected by these particular symptoms, which reveals gender-specific disparities. Wolfe-Christensen et al. (2017) also report differences in experiencing the diagnosis, mothers reached significantly higher levels of depressive symptoms than fathers. But these findings apply to parents of children with DSD in general and are not

specific for parents of children born with CAH. All in all, further research on the HrQoL of parents of CAH diagnosed children is needed.

2.4.2 Coping Strategies

Coping is a multi-faceted concept that is important in the context of psychosocial needs and adaptions in parents of children born with CAH. The originators of the transactional stress model Lazarus and Folkman (1984, p. 141) describe coping as ”[…] constantly changing cognitive and behavioural efforts to manage specific external and/or internal demands that are appraised as taxing or exceeding the resources of the person“. According to this theory, these efforts further can be problem-focused or emotion-focused and appear as a multitude of different coping mechanisms (Smith et al., 2016, Lazarus and Folkman, 1984). The outcome of the use of a particular coping mechanism is individual and dependent on many essential factors, e.g. the personal resilience of an individual (Smith et al., 2016). As Lazarus (2006, p. 111) states: ”No universally effective or ineffective coping strategy exists“. That is why it seems to be essential to measure the effectiveness of the individual coping behaviour of persons to consider their adaption to a problematic situation. According to Smith et al. (2016) the application of effective coping mechanisms leads to a decrease of psychological strain and therefore to an improved adaption of the situation.

Concerning the coping behaviour of parents of a child with CAH, there only exist few studies examining this subject particularly. As seen in the table below *(Table 2),* a coping mechanism, recurrently emphasized by affected parents, is getting into contact with other families with a CAH diagnosed child via support groups or other ways of communication (Barg et al., 2010, Boyse et al., 2014, Lundberg et al., 2017, Witt et al., 2018). Another practical aspect of coping with the new situation is to receive clear information about the disease pattern (Boyse et al., 2014, Lundberg et al., 2017, Witt et al., 2018). Affected families emphasized the importance of mediating disease information in an understandable and non-medical language (Boyse et al., 2014). Additionally, Witt et al. (2018) point out that the initial strain of facing the diagnosis lowered with gaining of information and a passing time.

Furthermore, parents seek for practical support for dealing with the everyday life challenges of caring for a child with CAH (Boyse et al., 2014, Lundberg et al., 2017). As Fleming et al. (2017a) show, provider instruction has a significant effect on the

well-being of a family dealing with CAH. They found out that the impact of the illness on the family lowers with an increasing parental care management ability. Furthermore, the findings of Witt et al. (2018), Lundberg et al. (2017), and Boyse et al. (2014) indicate professional psychosocial support being advisable to initiate, especially in the first years of the child's life. Aside from putting out often as helpful mentioned coping mechanisms, only Waldthausen (2007) measured the effectiveness of parental coping in CAH affected families. She found out that the applied coping mechanisms were above average effective in the surveyed parents (Waldthausen, 2007). However, further studies are necessary to validate these findings.

2.4.3 Special Needs

Regarding the primary needs of parents of CAH diagnosed children, most of the published studies show a high demand of psychosocial support (Barg et al., 2010, Bennecke et al., 2015, Simpson et al., 2018, Witt et al., 2018) as well as a need for more understandable information about the disease pattern of CAH (Boyse et al., 2014, Lundberg et al., 2017, Witt et al., 2018). The findings of Bennecke et al. (2015) exhibit a particular need for psychological support in 33.9% of the surveyed parents of children with CAH. This need increases with the amount of medical treatment a child gets. However, only a few parents in this study received a satisfying provision of psychological support (Bennecke et al., 2015). A study from Poland examining the mental and social problems of families affected by CAH shows similar findings; the surveyed parents addressed a shortage of satisfying psychological support (Barg et al., 2010).

Concerning the informational needs, parents wish to be informed in a simple way using non-medical language as well as oral and written information (Boyse et al., 2014, Lundberg et al., 2017). Lundberg et al. (2017) found that some parents had to deal with a lot of confusion after initially learning of the disease pattern CAH. The results of Boyse et al. (2014) show that especially the first information given about CAH left the parents overwhelmed and profoundly moved. They often experienced strong emotions and had "[…] a limited ability to absorb the information provided" (Boyse et al., 2014, p. 438). The pilot study of Witt et al. (2018) reveals that parents of CAH diseased children experienced a lot of intense feelings like shock, fear, grief, and despair while facing the diagnosis of their new-born. However, parents reported

that they were able to process the given information sometime later (Witt et al., 2018).

Good knowledge about CAH, though seems to be very important since Joshi et al. (2017) discovered a negative correlation between the knowledge about CAH and stress among the concerned parents. In addition, Fleming et al. (2017a) reported that parents who have been instructed on adrenal crisis management, have better management abilities which minimize the impact of CAH on the family. These findings highlight the importance of sufficient and accessible information after declaring that the new-born could be diseased with an illness like CAH.

Regarding this topic, some studies point out the parental need for more practical support. Parents require help with the handling of the medical conditions of their child in their everyday life, concerning e.g. the management of the child's medication or the processing of emergencies (Boyse et al., 2014, Lundberg et al., 2017, Simpson et al., 2018, Witt et al., 2018). Similar to other studies (Barg et al., 2010, Boyse et al., 2014, Witt et al., 2018) Simpson et al. (2018) emphasize the importance of parent-to-parent support for families with children with CAH or adrenal insufficiency. Having contact with other affected families constitutes a "[…] pivotal – source of support […]" (Simpson et al., 2018, p. 1452). Moreover, parents accentuated that they had to fight for a final and definite diagnosis, and 39% struggled to get access to a specialist with the appropriate knowledge and expertise of CAH, especially in Germany (Simpson et al., 2018).

Altogether, the specific needs of parents of children diagnosed with CAH seem to be very intense and broad *(Table 3)*. Further research is needed to confirm and/or widen these findings.

In conclusion, the parental HrQoL, coping behaviour, and satisfied needs are crucial for the child's development. According to Grootenhuis and Bronner (2009, p. 940) "Parent and child psychological reactions are significantly correlated […]". Epifanio et al. (2015) described a link between parental psychological condition and psycho-physical development of the child. Consequently, it is imperative to improve the situation of parents caring for a child diagnosed with CAH, because as Grootenhuis and Bronner (2009, pp. 940-941) declare: "[…] the better the parent is able to deal with the burdens of care, the better the child will be able to cope with his/her limitations, isolation and discomforts".

	Population	Intervention	Comparison	Outcome
Psychological Adjustment of Parents of Children Born with Atypical Genitalia 1 Year after Genitoplasty *(2017)* *Ellens et al.*	45 parents (♀25; ♂20) from the USA of 25 children (♀14; ♂11, age: 17 - 41 months, mean age: 26 months) diseased with DSD and/or atypical genitalia	questionnaires 1 year after surgical adjustment of the outer genitalia of the child (genitoplasty): demographic variables, BDI-II, BAI, PPUS, IES-R, SF-36, DRS	Suorsa et al. (2015), Wolfe-Christensen et al. (2017), subgroup analysis	- significant improvements in all psychological distress variables - clinically relevant distress in a subset of parents - parental mental health QoL significantly improved, parental physical QoL remained unchanged - 28% of parents showed some level of decisional regret
Changes in levels of parental distress after their child with atypical genitalia undergoes genitoplasty. *(2017)* *Wolfe-Christensen et al.*	49 parents (♀27; ♂22) from the USA of 28 children (♀17; ♂11) diseased with DSD and/or atypical genitalia	questionnaires 6 months after surgical adjustment of the outer genitalia of the child (genitoplasty): demographic variables, BDI-II, BAI, PPUS, IES-R, SF-36	Suorsa et al. (2015), published references, subgroup analysis	- no significant changes in level of depressive or anxious symptoms or QoL - lower QoL than published references - mothers reported significantly higher levels of depressive symptoms than fathers - significant decrease in level of posttraumatic stress symptoms 6-month post-operative
Characterizing Early Psychosocial Functioning of Parents of Children with Moderate to Severe Genital Ambiguity due to Disorders of Sex Development *(2015)* *Suorsa et al.*	51 parents (♀53%; ♂45%) from the USA of children (♀46%; ♂48%, ⚥ 4%, age: 6 - 23 months, mean age: 10 months) diseased with DSD and/or atypical genitalia	questionnaires before surgical adjustment of the outer genitalia of the child (genitoplasty): demographic variables, BDI-II, BAI, PPUS, IES-R, SF-36, Cosmetic Appearance Rating Scale	subgroup analysis	- dissatisfaction with the genital appearance of their child in 54.5% of questioned parents - more than 25% of parents reported experiencing a significant negative impact on QoL related to their mental health - a small but significant percentage of parents reported symptoms of anxiety, depression, uncertainty and posttraumatic stress - few gender differences
Die psychosoziale Situation von Familien mit einem an adrenogenitalem Syndrom erkrankten Kind *(2007)* *Waldthausen* *- unpublished doctoral thesis*	24 families from Germany of 24children (♀ 12; ♂ 12, (age: 10 months - 9.5 years, mean age: 3.9 years) diseased with CAH	questionnaires: FLZ, ULQIE, CHIP interview: clinical and sociodemographic variables, disease specific strains of the parents	published references, subgroup analysis	- high parental strain especially in the time facing the diagnosis - average - above-average life satisfaction - slight reduction in HrQoL - no gender or education differences in HrQoL - higher HrQoL in parents of a girl than parents of a boy - above average effectively coping

Abbreviations:
BAI The Beck Anxiety Inventory
BDI-II Beck Depression Inventory – 2nd Edition
CAH congenital adrenal hyperplasia
CHIP Coping Health Inventory for Parents
DRS Decisional Regret Scale
DSD Disorder of Sex Development
FLZ Fragebogen zur Lebens-zufriedenheit
HrQoL health-related quality of life
IES-R Impact of Event Scale Revised
PPUS Parental Perceptions of Uncertainty Scale
QoL quality of life
SF-36 Short Form health survey (36items)
ULQIE Ulmer Lebensqualitäts-Inventar für Eltern chronisch kranker Kinder.

	Population	Intervention	Comparison	Outcome
Psychosocial Situation of Parents with Children with Diagnosed 21-Hydroxylase Adrenogenital Syndrome (AGS): Preliminary Results of a Pilot Study *(2018)* *Witt et al.*	8 parents from Germany of 8 children (♀ 5; ♂ 3, age: 2 - 27 months, median age: 15 months) diseased with CAH	questionnaires: - general family characteristics: FAD - perceived burden: EBI - self-efficacy expectation: GSES - social support: OSS-3 open questions: - reaction to diagnosis - information and support needs	-	- increased burden and perceived disease-related stress of the affected parents, especially at the time of diagnosis - shortly after diagnosis parents reported a high need of social-, practical- and information support, which lowered within the course of time and gaining of information - especially helpful: contact with other affected families
From Knowing Nothing to Knowing What, How and Now: Parents' Experiences of Caring for their Children with Congenital Adrenal Hyperplasia. *(2017)* *Lundberg et al.*	20 parents (♀ 16; ♂ 4) from Sweden and the U.K. of 22 children (♀ 16; ♂ 6, age: 1 - 20 years, mean age: 9 years) diseased with CAH	semi-structured interviews: - general description of the child - parents' experiences of receiving the diagnosis - parents' and child's perceptions of health care and treatment - exchange with others - thoughts and questions about the child's future	-	parents emphasized the importance of knowing: - what is CAH? - what support needs my child? - coping strategies - how do I attend to my child's medical needs? - how do I talk to my child? parents reported challenges with: - connecting with the social network - emergency care - helping the child become independent parents characterized as helpful: - a good health communication - practical support - talking to a psychosocial professional - support groups/contact with other affected parents
"It was an overwhelming thing": parents' needs after infant diagnosis with congenital adrenal hyperplasia *(2014)* *Boyse et al.*	6 parents (♀ 4; ♂ 2) of 4 children (♀ 2; ♂ 2, age: 5 - 11 years) diseased with CAH	semi-structured telephone interviews: - How do parents learn about CAH and its management? - How do parental needs could have been better met during the time of first diagnosis?	-	parents emphasized the importance of: - better health communication - decision support - practical support - parent-to-parent social support

Abbreviations: CAH congenital adrenal hyperplasia; EBI Eltern-Belastungs-Inventar; FAD Family Assessment Device; GSES Generalized Self-Efficacy Scale; OSS-3 Oslo 3-items Social Support Scale.

	Population	Intervention	Comparison	Outcome
The analysis of psychological and social problems, the physical development in young people with congenital adrenal hyperplasia - owner experience *(2010)* *Barg et al.*	parents from Poland of 20 children (♀ 7; ♂ 13, age: 4.5 - 19 years) diseased with CAH	questionnaires: - child's development - emotional reactions to diagnosis - concerns and expectations - knowledge about CAH - gender assignment of the child	-	- emotional reaction to diagnosis: 60% anxiety, 40% helplessness - concerns: 40% worries about the further sexual life of the child - most of the parents were satisfied with the way they were informed about CAH, but 15% didn't fully understand the matter of this disorder - > 60% searched for additional information about CAH - parents emphasized the importance of support groups for children with CAH - parents complained about lack of satisfying psychological support
Die psychosoziale Situation von Familien mit einem an adrenogenitalem Syndrom erkrankten Kind *(2007)* *Waldthausen* *- unpublished doctoral thesis*	24 families from Germany of 24 children (♀ 12; ♂ 12, age: 10 months - 9.5 years, mean age: 3.9 years) diseased with CAH	questionnaires: - life satisfaction: FLZ - HrQoL: ULQIE - coping: CHIP interviews: - clinical and sociodemographic variables - disease specific strains of the parents	published references, subgroup analysis	- high parental strain especially in the time facing the diagnosis - average - above-average life satisfaction - slight reduction in HrQoL - no gender or education differences in HrQoL - parents of girls had a higher HrQoL than parents of boys - above average effectively coping

Abbreviations: CAH congenital adrenal hyperplasia; CHIP Coping Health Inventory for Parents; FLZ Fragebogen zur Lebenszufriedenheit; HrQoL health-related quality of life; ULQIE Ulmer Lebensqualitäts-Inventar für Eltern chronisch kranker Kinder.

Population		Intervention	Comparison	Outcome
Psychosocial Situation of Parents with Children with Diagnosed 21-Hydroxylase Adrenogenital Syndrome (AGS): Preliminary Results of a Pilot Study *(2018)* *Witt et al.*	8 parents from Germany of 8 children (♀ 5; ♂ 3, age: 2 - 27 months, median age: 15 months) diseased with CAH	questionnaires: - general family characteristics: FAD - perceived burden: EBI - self-efficacy expectation: GSES - social support: OSS-3 open questions: - reaction to diagnosis - information and support needs	-	- increased burden and perceived disease-related stress of the affected parents, especially at the time of diagnosi - shortly after diagnosis parents report a high need of social-, practical- and information support, which lowered within the course of time and gaining of information - especially helpful: contact with other affected families
Adrenal Insufficiency in Young Children: a Mixed Methods Study of Parents' Experiences *(2018)* *Simpson et al.*	parents of children diseased with CAH or AI from the U.K., the Netherlands, Germany a) 57 parents (♀43; ♂14) of 57 children (♀25; ♂31; ⚥ 1, age: ≤ 6 years) b) 20 parents (♀14; ♂6) of 17 children (♀9; ♂8, age: 3 months - 10 years)	a) online survey b) semi-structured interviews subjects: diagnosis, treatment, impact, information, support, hopes and concerns for the future	comparison among each other, comparison by country	parents accentuate: - had to fight for a diagnosis - latent anxiety because of the medication - skills, knowledge, confidence developed over time - worried about the child's future, the transition from paediatric to adult health services - importance of contact with other affected families - unmet needs for practical and emotional support (due to lack of knowledge of the condition amongst local health teams, lack of access to psychological services) 76% left hospital well informed 39% struggled to get access to specialists with appropriate knowledge/expertise (especially in Germany)
From Knowing Nothing to Knowing What, How and Now: Parents' Experiences of Caring for their Children With Congenital Adrenal Hyperplasia. *(2017)* *Lundberg et al.*	20 parents (♀ 16; ♂ 4) from Sweden and the U.K. of 22 children (♀ 16; ♂ 6, age: 1 - 20 years, mean age: 9 years) diseased with CAH	semi-structured interviews: - general description of the child - parent's experiences of receiving the diagnosis - parent's and child's perceptions of health care and treatment - exchange with others thoughts and questions about the child's future	-	parents emphasized the importance of knowing: - what is CAH? - what support needs my child? - coping strategies - how do I attend to my child's medical needs? - how do I talk to my child? parents reported challenges with: - connecting with the social network - emergency care - helping the child become independent parents characterize as helpful: - a good health communication - practical support - talking to a psychosocial professional - support groups / contact with other affected parents

Abbreviations: AI adrenal insufficiency; CAH congenital adrenal hyperplasia; EBI Eltern-Blastungs-Inventar; FAD Family Assessment Device; GSES Generalized Self-Efficacy Scale; OSS-3 Oslo 3-items Social Support Scale.

	Population	Intervention	Comparison	Outcome
Subjective need for psychological support (PsySupp) in parents of children and adolescents with disorders of sex development *(2015)* *Bennecke et al.*	593 parents (♀ 315; ♂ 278) from Germany, Austria, Switzerland of 329 girls (mean age: 7.25 years) diseased with DSD, CAH proportionate 38.9%	questionnaires: - sociodemographic variables, medical history, child development, peer relations, questions concerning DSD - need for psychological support: DSD-questionnaire - child behaviour: CBAQ	comparison among each other (disease groups)	- need for psychological support: 40.4% in all parents 33.9% in parents of children with CAH - 50% of all parents received it adequately - need did not change with sex assignment surgery - need increased with the amount of medical treatment
"It was an overwhelming thing": parents' needs after infant diagnosis with congenital adrenal hyperplasia *(2014)* *Boyse et al.*	6 parents (♀ 4; ♂ 2) of 4 children (♀ 2; ♂ 2, age: 5 - 11 years) diseased with CAH	semi-structured telephone interviews: - How do parents learn about CAH and its management? - How do the parents' needs could have been better met during the time of first diagnosis?	-	parents emphasized the importance of: - better health communication - decision support - practical support - parent-to-parent social support
The analysis of psychological and social problems, the physical development in young people with congenital adrenal hyperplasia - owner experience *(2010)* *Barg et al.*	parents from Poland of 20 children (♀ 7; ♂ 13, age: 4.5 - 19 years) diseased with CAH	questionnaires: - child's development - emotional reaction to diagnosis - concerns and expectations - knowledge about CAH - gender assignment of the child	-	- emotional reaction to diagnosis: 60% anxiety, 40% helplessness - concerns: 40% worries about the further sexual life of the child - most of the parents were satisfied with the way they were informed about CAH, but 15% didn't fully understand the matter of this disorder - > 60% searched for additional information about CAH - parents emphasized the importance of support groups for children with CAH - parents complained of lack of satisfying psychological support

Abbreviations: CAH congenital adrenal hyperplasia CBAQ Child Behaviour and Attitudes Questionnaire DSD disorder of sex development.

3. Material and Methods

This doctoral thesis is based on a retrospective cross-sectional study examining the HrQoL, coping, and needs of parents caring for a child with CAH diagnosed by new-born screening. In a mixed-method approach, quantitative and qualitative data were analysed. With this method, an extensive and thorough overview of the psychosocial situation of affected parents is enabled. This doctoral thesis focuses on the quantitative data analysis, aiming to gain knowledge about HrQoL, coping, and the needs of parents of children born with CAH.

3.1 Participants

In a retrospective cross-sectional study, parents caring for a child with CAH diagnosed by new-born screening were asked to report on their HrQoL, coping strategies as well as their needs. The study was planned as a cooperation project of the Department of Medical Psychology of the University of Hamburg-Eppendorf [UKE], the Bavarian State Office for Health and Food Safety (Bayerisches Landesamt für Gesundheit und Lebensmittelsicherheit, Screeningzentrum Bayern [LGL]), and the University Children's Hospital in Erlangen. Overall, 120 families were recruited through the database of the LGL. The database refers to parents who participated in the population-based study "Langzeitverlauf der im Neugeborenenscreening entdeckten Kinder" (Odenwald et al., 2015, Odenwald et al., 2016) of the LGL. Respective children were born between 1999 and the first months of 2018. Addressing these mothers and fathers, 83.9% of all parents with a child diagnosed with CAH by new-born screening in Bavaria were included*.

Including criteria were: having a child with a 21-OH deficiency which was detected by new-born screening in Bavaria. From September 2018 until September 2019, a total of 104 families with one CAH diagnosed child and 16 families with two CAH diagnosed children were consulted. It is pursued to interrogate both, mothers and fathers. Excluding criteria were a missing declaration of consent, insufficient knowledge of German as well as other diseases than CAH being the centre of attention of the family.

* data provided by the LGL

3.2 Data Collection

The quantitative data collection included different questionnaires combined into one package for each mother and father. Data collection started after receiving the ethics approval for human research of the Bavarian ethic committee in September 2018 (No. 18003). Questionnaire packages were sent to eligible parents by the LGL via postal service including an informative letter about this study, a declaration of consent, and a pre-paid return envelope addressed to the UKE in Hamburg. Data analysis was confirmed at the UKE. One reminder was sent to the families two months after initially sending out the questionnaire packages. Based on self-report, parents provide information about their HrQoL in the last week, their coping strategies for dealing with the new situation due to their child's disease and their current particular needs. Additionally, parent-reported data about sociodemographics, the child's health condition and circumstances regarding the diagnosis process were collected.

The HrQoL of the parents was measured using the chronic-generic Ulmer Lebensqualitätsinventar für Eltern chronisch kranker Kinder [ULQIE] developed by Goldbeck and Storck (2002). This questionnaire consists of 29 items divided into five subscales examining the HrQoL of the participant within the last week. The subscales include the dimensions (1) *physical and daily functioning*, (2) *satisfaction with the situation in the family*, (3) *emotional distress*, (4) *self-development*, and (5) *well-being* as well as some items without any scale assignment. Each item is rated on a 5-point Likert-scale ranging from zero to four. The total score of the ULQIE is computed by adding the scores of the 19 positive expressed items to the recoded scores of the remaining ten negative expressed items. Proceeding like this, a maximum value of 116 points is possible, whereas half of the published norm, marked by the 25th and 75th percentile, score in a range of 59-83 points. The internal consistency (Cronbach's Alpha) for the subscales is between .75 and .88, and for the global scale .91 (Goldbeck and Storck, 2002). The ULQIE was used in multiple studies, e.g. the examinations of Waldthausen (2007), and West et al. (2009). Further information about these reference groups can be inspected under the headline *5. Discussion*. In this study, the calculated reliability of the questionnaire fulfils the scientific norms with an inner consistency (Cronbach's Alpha) between .74 and .86 for the subscales, and .92 for the total scale.

Parental coping strategies were assessed using the chronic-generic Coping Health Inventory for Parents [CHIP] questionnaire developed by McCubbin et al. in 1983 (McCubbin et al., 1983). The German version was validated by McCubbin et al. in 2001 (McCubbin et al., 2001). The CHIP consists of 45 items that examine the usefulness of different coping strategies of parents of chronically ill children in the present or the past. The items are assigned to three subscales: (1) *maintaining family integration, cooperation, and an optimistic definition of the situation*, (2) *maintaining social support, self-esteem, and psychological stability*, and (3) *understanding the medical situation through communication with other parents and consultation with medical staff*. All items are rated on a 4-point Likert-scale ranged from zero to three with an additional option to choose the answers 'did not try' or 'was not possible'. The total score of the CHIP is computed by adding up all item scores, whereas the additional response options count as zero. Overall, a maximum total score of 135 is possible. Half of the published norm reach scores between 64-89 points, considering the cohort between the 25th and 75th percentile. Cronbach's Alpha for the subscales is between .71 and .76, and for the global scale .86 (McCubbin et al., 2001). Some studies applied the CHIP, e.g. the examinations of Senger et al. (2016) and Clever et al. (2019). Further information about these reference groups can be inspected under the headline *5. Discussion*.

Chronic-generic needs of parents were measured using the Bedürfnis-Skala für Eltern chronisch kranker Kinder. This questionnaire contains 19 items about the parental need of information, psychosocial care, exchange with parents affected the same way and support in everyday life. It is part of an unpublished habilitation treatise from Dr. phil. Wiedebusch-Quante. The items are rated on a 5-point Likert-scale ranging from one to five. The higher the score of one item, the more intense the need of the parent. The reliability of the Bedürfnis-Skala für Eltern chronisch kranker Kinder fulfils the scientific norms with an inner consistency (Cronbach's α) of .94 (Wiedebusch-Quante, 2009). Beyond that, it was used in a doctoral thesis (Schreiber, 2011). Further information about these reference groups can be inspected under the headline *5. Discussion*.

3.3 Data Analysis

Quantitative data from the questionnaire package were collected anonymously in an Excel sheet and analysed using the statistic software SPSS, version 25 (IBM, 2017).

After conducting a plausibility test initially, descriptive statistics were computed for each subscale of the questionnaires examining HrQoL and coping. Afterwards, the global scales of the ULQIE, CHIP, and Bedürfnis-Skala für Eltern chronisch kranker Kinder were calculated to gain an overview of the psychosocial situation of parents caring for a child with CAH. Missing values were replaced by the individual mean score for each variable if missing data were random or less than 20% of all scale items (Hawthorne and Elliott, 2005). The scores of mothers and fathers were compared to each other and published cohorts of parents whose children suffer from different chronic diseases. The comparison with reference data was carried out by using Student's t-tests. Multiple linear regression analyses were performed to identify variables that predict parental HrQoL. Sociodemographic and clinical variables were entered as predictors into the model, as well as the CHIP total score and the total score of the Bedürfnis-Skala für Eltern chronisch kranker Kinder. The clinical data comprise the child's age and sex and whether or not the child is affected by life-threatening salt-wasting crises by registering the medication intake of Fludrocortisone. The examined sociodemographic aspects consist of the parents' age and sex, the number of children in general, the number of children diseased with CAH, the place of residence, the educational qualification, and the employment status in the last 12 months. Further examined aspects are the use of psychosocial consultation and contact with other parents who have children with CAH.

For a confirmatory approach, a Bonferroni-adjustment for multiple testing was conducted. Based on an initial significance level of $\alpha = .05$, the significance level was reduced to .0033. The reduction is justified by simultaneously calculating multiple linear regression analyses and one one-sample t-test. The linear regression model investigates 14 relevant influencing factors to analyse the influence on the HrQoL measured by the ULQIE total value. With the one-sample t-test, the computed ULQIE total value was compared to a reference cohort's total value.

In conclusion, exploratory regression analyses were made to investigate the impact of clinical or sociodemographic aspects on the CHIP's total score, on the total score of the Bedürfnis-Skala für Eltern chronisch kranker Kinder, as well as on each

subscale of the ULQIE. Besides, it was examined exploratory if the coping behaviour or parents' intensity of needs influence the ULQIE-subscales.

4. Results

With 105 letters of return out of originally 240 letters sent, the overall response rate was 43.8% *(Figure 1)*.

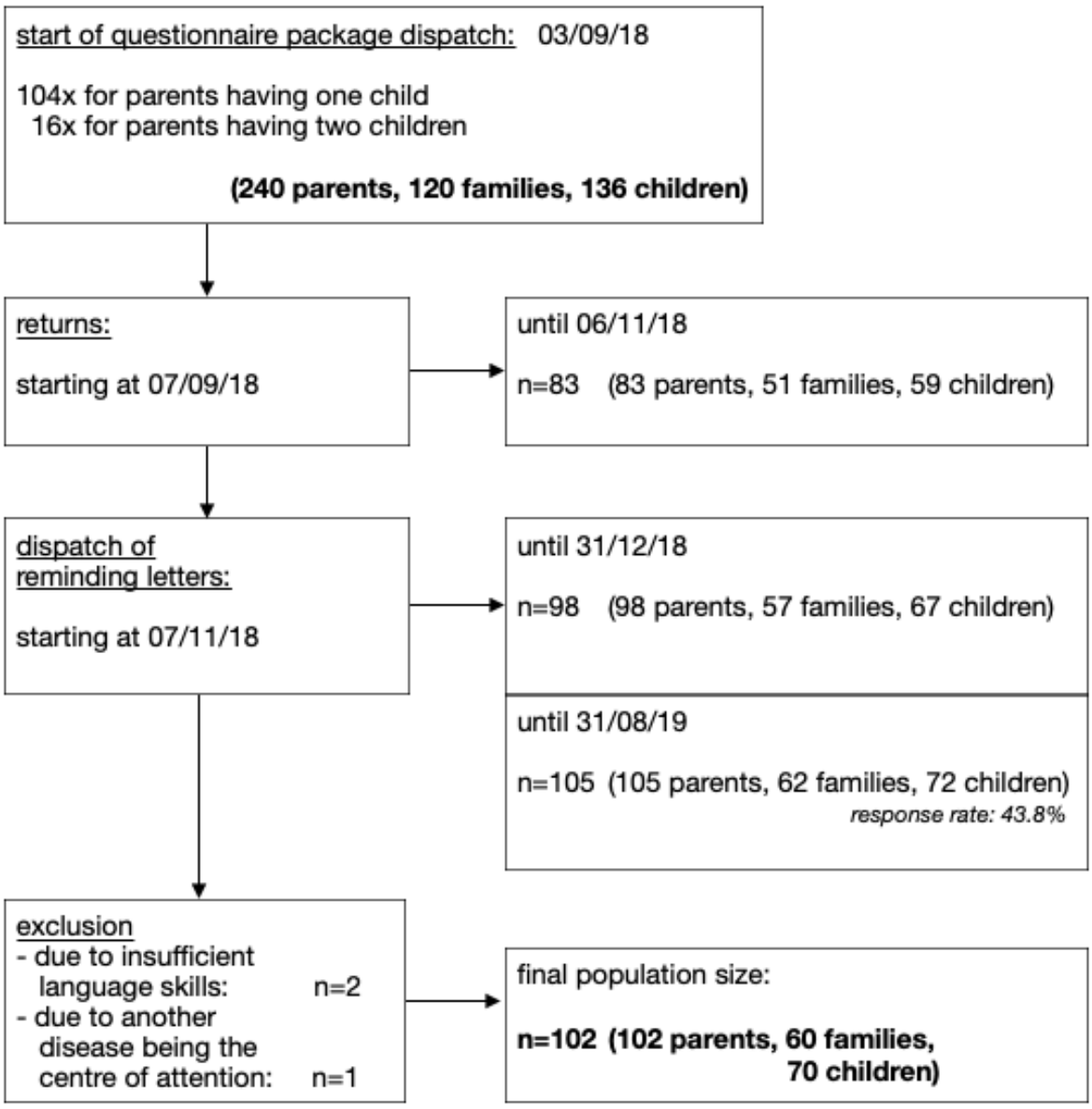

Figure 1: Algorithm of Data Collection (09/2018-09/2019)

4.1 Sample Characteristics

Among the 105 parents with CAH diagnosed children who filled out the questionnaire, three were excluded due to one or more of the exclusion criteria. In one family, another disease than CAH was the focus of concern. Corresponding data must be excluded, since another disease may influence the answers. Besides, two respondents had to be excluded due to insufficient language skills *(Figure 1)*.
In total, 60 families were included in the analyses, consisting of 102 parents (60 mothers and 42 fathers). In 42 cases, both parents of one family answered the questionnaires, and 18 times only one parent per child sent back data. Ten of the 60 families each have two children with CAH, including two families giving birth to CAH affected twins. In conclusion, parents of 70 children diagnosed with CAH participated. Further detailed information concerning parents' and children's characteristics, as well as detailed information regarding the time of diagnosis, are listed in *tables* *Table* 4-*Table* 6.

Table 4: Parents' Characteristics (n=102)

Age (in years)	M ± SD	43.3 ± 7.4
	min	25.0
	max	61.0
Gender (n; %)	female	60 (58.8)
	male	42 (41.2)
Marital status (n; %)	single parent	6 (5.9)
	partnership	93 (91.2)
	missing	3 (2.9)
Place of residence (n; %)	large town [a]	13 (12.7)
	middle town [b]	19 (18.6)
	small town [c]	16 (15.7)
	countryside [d]	53 (52.0)
	missing	1 (1.0)
Educational qualifications (n; %)	low [e]	29 (28.4)
	middle [f]	36 (35.3)
	high [g]	32 (31.4)
	other	5 (4.9)
Employment (n; %)	full-time job	50 (49.0)
	part-time job	36 (35.3)
	parental leave	7 (6.9)
	other	8 (7.8)
	missing	1 (1.0)

[a] ≥ 100.000 residents
[b] ≥ 20.000 – 99.999 residents
[c] ≥ 5.000 – 19.999 residents
[d] < 5.000 residents
[e] no qualification or German "Hauptschulabschluss" (corresponds to certificate of secondary education)
[f] German "mittlere Reife" or "Abitur" (corresponds to ordinary level or A-level)
[g] bachelor's or master's degree

M: arithmetic mean value, max: maximum value, min: minimum value, SD: standard deviation.

Table 5: Children's Characteristics (n=70)

Age (in years)	M ± SD	11.1 ± 4.9
	Median	11.6
	min	.7
	max	19.4
Gender (n; %)	female	32 (45.7)
	male	38 (54.3)
Complications after birth (n; %)	none	44 (62.9)
	with the mother	3 (4.3)
	with the child	20 (28.6)
	with both	3 (4.3)
Therapy [a] (n; %)	Hydrocortisone	70 (100)
	Fludrocortisone	57 (81.4)
	NaCl solution	23 (32.9)
Hospital admission due to adrenal crisis / due to hypoglycaemia (n; %)	no	41 (58.6)
	yes, once	13 (18.6)
	yes, repeatedly	15 (21.4)
	missing	1 (1.4)
Genital ambiguity of girls at birth (n; %)	no	9 (28.1)
	yes	23 (71.9)
Prior surgeries (genitoplasty) of girls with genital ambiguity (n; %)	no	1 (4.3)
	yes	22 (95.7)
– if yes, age at surgery (in months)	M ± SD	14.8 ± 18.8
	Median	11.5
	min	2
	max	96

[a] multiple responses possible

M: arithmetic mean value, max: maximum value, min: minimum value, NaCl: sodium chloride, SD: standard deviation.

Table 6: Communicating the Diagnosis (n=102)

Age of the child at presumptive diagnosis (in days)	M ± SD	8.2 ± 7.5
	Median	6.5
	min	.0
	max	35.0
Way of transferring the presumptive diagnosis (n; %)	via direct conversation	63 (61.8)
	via telephone call	36 (35.3)
	other	3 (2.9)
Bearer of the presumptive diagnosis (n; %)	obstetrician	1 (1.0)
	nurse	4 (3.9)
	physician at the maternity clinic	48 (47.1)
	paediatrician	27 (26.5)
	other	17 (16.7)
	missing	5 (4.9)
Receipt of psychosocial support at the ward (n; %)	no	71 (69.6)
	yes	13 (12.7)
	missing	18 (17.6)
Bearer of the final diagnosis (n; %)	paediatric endocrinologist	38 (37.3)
	ward physician	17 (16.7)
	assistant medical director or head doctor	40 (39.2)
	other	2 (2.0)
	missing	5 (4.9)
Sufficient information about the disease (n; %)	no	24 (23.5)
	yes	76 (74.5)
	missing	2 (2.0)

Sources of information [a] (n; %)	internet	48 (47.1)
	physician	70 (68.6)
	specialist books	4 (3.9)
	other	19 (18.6)
	none	3 (2.9)
	missing	8 (7.8)
Contact with other parents [a] (n; %)	self-help group	30 (29.4)
	internet forums	8 (7.8)
	social networks	6 (5.9)
	other	6 (5.9)
	none	57 (55.9)
	missing	6 (5.9)
Utilization of psychosocial consultation (n; %)	no	86 (84.3)
	yes	16 (15.7)
- Was it helpful? (n; %)	not at all	2 (12.5)
	a little / partly	4 (25.0)
	much / a lot	10 (62.5)
Age of the child at first out-patient visit to a paediatric endocrinologist (in days)	M ± SD	67.8 ±119.9
	Median	40.0
	min	7.0
	max	1095.0
	missing (n; %)	8 (7.8)

[a] multiple responses possible

CAH: congenital adrenal hyperplasia, M: arithmetic mean value, max: maximum value, min: minimum value, SD: standard deviation.

4.2 HrQoL of Parents with Children Born with CAH

Within this study, parents of children affected by CAH reported HrQoL scores using the ULQUIE. This questionnaire scored in the upper segment of the 0-4 scale, with the highest means for the subscale *satisfaction with the situation in the family* (3.37 ± .59) and the lowest means for the subscale *self-development* (2.30 ± .78) *(Figure 2)*.

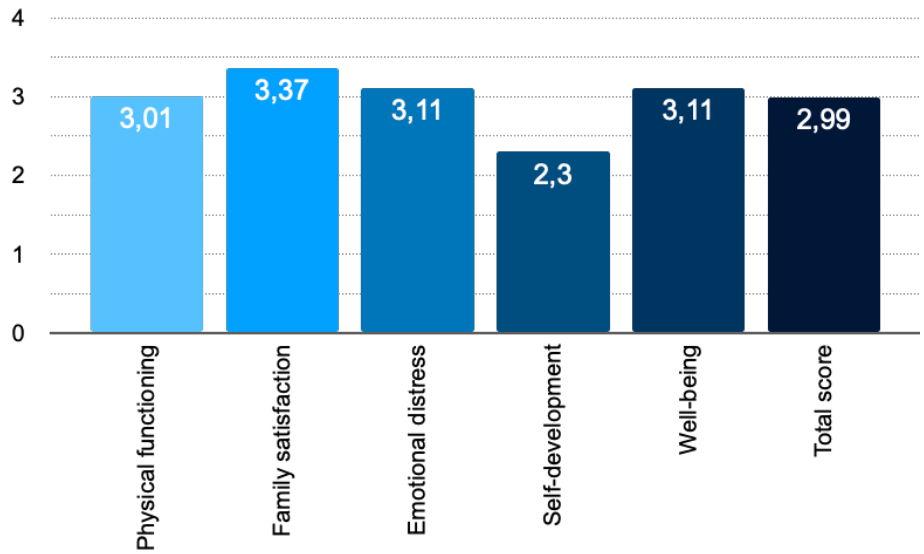

Figure 2: ULQIE - Results

In comparison to German reference values (Goldbeck and Storck, 2002), parents of CAH diagnosed children reported significantly higher HrQoL scores for *physical and daily functioning, emotional distress, self-development, well-being* as well as for the ULQUIE total score ($t(99)=8.46$, $p \leq .001$) *(Table 7)*. Thus, these parents mark the 84th percentile concerning the ULQIE total score, referring to percentile data from Goldbeck and Storck (2002). Further comparison with reference data can be seen in the table below *(Table 7)*.

Table 7: ULQIE - Comparison with Reference Data

Perspective	Domains	n	M	SD	Reference -values	t	df	p
Parent-reported HrQoL	Physical functioning	100	3.01	.62	2.44 [a]	9.24	99	**≤.001**
	Family satisfaction	98	3.37	.59	3.28 [a]	1.57	97	.119
	Emotional distress	100	3.11	.66	2.30 [a]	12.34	99	**≤.001**
	Self-development	100	2.30	.78	1.93 [a]	4.76	99	**≤.001**
	Well-being	101	3.11	.66	2.68 [a]	6.58	100	**≤.001**
	Total	100	2.99	.51	2.56 [a]	8.46	99	**≤.001**
Percentile data	**Total**	96	78	21.61	70 [b]	3.86	95	≤.001
	- mothers	56	77	24.57	55 [c]	6.96	55	≤.001
	- fathers	40	80	18.70	60 [c]	6.61	39	≤.001

a with reference to Goldbeck et al. (2002); confirmatory approach
b with reference to Waldthausen (2007); exploratory approach
c with reference to West et al. (2009); exploratory approach

df: degrees of freedom, HrQoL: health-related quality of life, M: arithmetic mean value, p: p-value, SD: standard deviation, t: t-value.

No meaningful differences between the HrQoL scores of mothers and fathers were found. The same applies for parents of one child with CAH in comparison to parents of two CAH affected children. However, the analyses showed differences for the ULQUIE total score between parents of younger (< 10 years) and older CAH children (≥ 10 years), as well as between parents of CAH girls and CAH boys *(Table 8)*. According to Sawyer et al. (2018), the age of ten marks the beginning of adolescence. Parents of children with CAH being in adolescence reach higher ULQIE total scores than parents of children aged younger than ten years. Further, parents with a girl with CAH seem to have a higher HrQoL than a boy's parents.

Table 8: ULQIE - Subgroup Analysis

Subgroups	n	M	SD
Mothers	58	2.99	.50
Fathers	42	2.99	.52
Parents of children < 10 years	32	2.95	.60
≥ 10 years	61	3.01	.44
Parents of girls	41	3.07	.53
boys	59	2.93	.48
Parents with 1 CAH child	83	2.99	.50
> 1 CAH children	17	2.98	.55

CAH: congenital adrenal hyperplasia, M: arithmetic mean value, SD: standard deviation.

The regression analysis contributes to explaining a substantial variance of parental HrQoL assessed using the ULQIE total score ($F(14,44)=1.829$, $p=.064$, $n=58$). The results show that the ULQIE total score was not significantly influenced by any sociodemographic or clinical particulars. Nonetheless, the examined HrQoL of the parents depends considerably on the effectiveness of applied coping strategies and the intensity of needs of these parents. Confirmatory regression analyses show a significant positive correlation between the total score of the CHIP and the ULQIE total value, which means that parents with highly developed coping strategies reached higher values on the measured HrQoL. Additionally, the data convey, that parents with intense needs achieve significantly lower scores on the measured HrQoL scale *(Table 9)*. The coefficient of determination R^2 for the linear regression model comes to .404.

Table 9: ULQIE - Influencing Factors on ULQIE total score

Influencing factors	CoR	SE	p	CI (lower endpoint|upper endpoint)
Constant term	2.21	.52	≤.001	1.17 | 3.25
Gender of the parent (being a mother)	- .03	.17	.843	- .38 | .31
Age of the parent	.02	.01	.137	- .01 | .04
Number of children in general	- .01	.07	.851	- .15 | .13
Number of children with CAH	- .09	.16	.564	- .41 | .23
Place of residence (countryside)	.05	.11	.654	- .18 | .28
Educational qualification (high)	- .00	.13	.973	- .26 | .25
Employment (part-time job)	.07	.17	.663	- .27 | .41
Psychosocial consultation (yes)	- .04	.19	.852	- .41 | .34
Contact with other parents (yes)	- .14	.12	.244	- .37 | .10
Gender of the child (having a girl)	.04	.13	.747	- .21 | .29
Age of the child	- .00	.02	.877	- .03 | .03
Phenotype of CAH (salt-wasting type)	- .17	.15	.278	- .48 | .14
CHIP total score	.55	.14	**≤.001**	.27 | .83
Bedürfnis-Skala total score	- .26	.08	**.002**	- .42 | .11

CAH: congenital adrenal hyperplasia, CHIP: Coping Health Inventory for Parents, CI: confidence interval, CoR: coefficient of regression, p: p-value, SE: standard error, ULQIE: Ulmer Lebensqualitätsinventar für Eltern chronisch kranker Kinder.

Exploratory regression analyses show a correlation between particular subscales of the ULQIE, and several factors involved. The subscales *daily functioning*, *family satisfaction*, and *emotional distress* seem to depend on coping strategies and needs in the same way as the total score of the ULQIE does *(Table 10)*. The HrQoL measured by the subscale *family satisfaction* is further influenced by the age of the child (*B*=-.05, *p*=.009) and the use of psychosocial consultation (*B*=-.46, *p*=.046). Parents who used any kind of psychological or social-pedagogic help seem to have a lower HrQoL, measured by the *family satisfaction* subscale. The same applies to parents with an older child with CAH. Concerning the subscale *self-development* regression analyses show impacts from the parents' educational qualification, the gender of their child, and their applied coping strategies. The higher the degree, the lower seem to be the HrQoL measured by the subscale *self-development* (*B*=-.51, *p*=.031). Additionally, the HrQoL on the *self-development* scale of parents of a boy decreases about .44 units compared to parents of a girl (*p*=.061). The coping strategies influence the *parents' self-development* in the same way as the total score is influenced *(Table 10)*. The subscale *well-being* produces relatively high p-values.

Therefore, no well-founded tendencies can be suggested. Nevertheless, none of these findings could be proven in a significant manner due to an exploratory approach.

Table 10: ULQIE Subscales - Influencing Factors

Influencing factors on ULQIE subscales	**daily functioning** (CoR | p)	**family satisfaction** (CoR | p)	**emotional distress** (CoR | p)	**self-development** (CoR | p)	**well-being** (CoR | p)
CHIP [a]	.43 | .024	.71 | ≤.001	.40 | .068	.86 | .001	.37 | .088
Bedürfnis-Skala [a]	-.24 | .026	-.30 | .003	-.35 | .005	-.23 | .118	-.14 | .233

a total score
CHIP: Coping Health Inventory for Parents, CoR: coefficient of regression, p: p-value, ULQIE: Ulmer Lebensqualitätsinventar für Eltern chronisch kranker Kinder.

4.3 Coping of Parents with Children Born with CAH

The effectiveness of the parents' individual coping behaviour was measured using the CHIP. Mothers and fathers in this study stated *maintaining family integration, cooperation, and an optimistic definition of the situation* as their most helpful coping pattern (2.31 ± .45). *Maintaining social support, self-esteem, and psychological stability* was deemed least helpful compared to the other subscales. Nonetheless, this subscale scores in the upper-middle segment of the 0-3 scale (1.76 ± .60). With a mean total score of 2.02 (± .47), the effectiveness of coping mechanisms of parents of children with CAH is comparable to German reference cohorts (McCubbin et al., 2001, Clever et al., 2019). Mothers in this study rank the helpfulness of particular coping patterns on average higher than fathers *(Table 11)*.

Table 11: CHIP - Results

CHIP Domain		**M**	**SD**
Subscales	(1) Family integration	2.31	.45
	(2) Social support	1.76	.60
	(3) Medical knowledge	1.93	.69
Total		2.02	.47
	- mothers	2.11	.41
	- fathers	1.90	.52

CHIP: Coping Health Inventory for Parents, M: arithmetic mean value, SD: standard deviation.

The results reveal that the CHIP total score was not influenced by any sociodemographic or clinical particulars. The exploratory regression analyses indicate relatively high p-values on every influencing factor ($p \geq .094$).

4.4 Needs of Parents with Children Born with CAH

With the Bedürfnis-Skala für Eltern chronisch kranker Kinder, the intensity of needs of parents with CAH diagnosed children was measured. On a 1-5 scale, parents set their mean intensity of needs at 2.81 (±.80). Comparing mothers and fathers, the mean total scores differ about .13 points, resulting from mothers mean score of 2.86 and a mean score of 2.73 for fathers. The most frequently indicated needs were primarily informational, like the need for more information about therapy options, dealing with departments and councils, diagnostic methods, and dealing with the ill child. Another often mentioned requirement was the need to have more time for the partner. The three needs least indicated are the need to get more relief in everyday life from relatives or friends and conducting problem-centred conversations with friends *(Table 12)*.

Table 12: Bedürfnis-Skala - Results

Perspective	Items	n	min	max	M	SD
Bedürfnis-Skala für Eltern chronisch kranker Kinder	Information about therapy options	98	1	5	4.01	1.14
	Information about dealing with departments and councils	93	1	5	3.47	1.40
	Information about diagnostic methods	90	1	5	3.46	1.38
	Having more time for my partner	88	1	5	3.44	1.06
	Information about dealing with my ill child	93	1	5	3.34	1.43
	Having more time for oneself	90	1	5	3.07	1.17
	Speak more about my problems with my partner	84	1	5	3.02	1.41
	Meeting up with other affected parents	89	1	5	2.87	1.36
	Having more time for other family members	89	1	5	2.85	1.15
	Having more time for my friends	89	1	5	2.65	1.04
	Having more time for my other child(ren)	71	1	5	2.65	1.52
	Information about dealing with my other child(ren)	77	1	5	2.58	1.47

	Speak more about my problems with other affected parents	87	1	5	2.55	1.27
	Getting more relief in everyday life from my partner	84	1	5	2.54	1.24
	Speak more about my problems with relatives	87	1	5	2.47	1.24
	Having a contact in the treatment centre to speak with about my problems (psychologist/social education worker)	88	1	5	2.31	1.24
	Getting more relief in everyday life from relatives	85	1	5	2.19	1.08
	Speak more about my problems with friends	89	1	5	2.18	1.04
	Getting more relief in everyday life f rom friends	85	1	5	1.80	0.96
	Total	**82**	**1**	**4.37**	**2.81**	**0.80**

M: arithmetic mean value, max: maximum value, min: minimum value, SD: standard deviation.

The mean total score of the Bedürfnisskala für Eltern chronisch kranker Kinder in this study's sample is comparable to German reference data (Wiedebusch-Quante, 2009, Schreiber, 2011). Nevertheless, slight differences were found in two of the three diagnose subgroups examined by Wiedebusch-Quante (2009). Parents of children with haemophilia (2.56 ± .70) show lower needs, and parents of children with juvenile idiopathic arthritis state higher needs (3,02 ± .71) (Wiedebusch-Quante, 2009) than the parents in the present sample.

Further, parent-reported needs showed differences depending on the child's gender. Exploratory regression analyses show that parents of a boy declare their needs more severely than parents giving birth to a girl with .67 units difference (p=.004). Other factors included into the model did not show any substantially dependences.

5. Discussion

The study aimed to investigate the psychosocial situation of parents of children with CAH who were diagnosed by new-born screening. Several aspects have been assessed, including HrQoL, coping strategies, and needs of 102 parents using the ULQIE, the CHIP, and the Bedürfnis-Skala für Eltern chronisch kranker Kinder.

5.1 General Considerations

In this study, parents reported an above-average HrQoL compared to reference data. The comparison was drawn on the basis of percentile data by Goldbeck and Stork. In 2002, the authors published a study where 244 parents of children diagnosed with an oncological disease, diabetes mellitus, or epilepsy were surveyed. The children of these parents were on average younger than the children in the present study. Additionally, the duration between the initial diagnoses and the survey showed with ten to 14 weeks shorter intervals in Goldbeck and Stork's sample (Goldbeck and Storck, 2002). The comparison to other study populations revealed similar results. West et al. (2009) investigated the HrQoL of 386 parents of children suffering from an oncological or cardiological disease or from cystic fibrosis. The mean time since diagnosis was about four years for this population. The parents reached a lower mean total score using the ULQIE than the mothers and fathers in the present study (West et al., 2009). The same applies to a study from 2007, where Waldthausen examined 41 parents whose children were diagnosed with CAH. The mean age of these children was 3.9 years, and so was the meantime since diagnosis. The parents examined by Waldthausen reached lower scores on the ULQIE than the mothers and fathers in the present study. Both cohorts differ in the time since diagnosis of about seven years (Waldthausen, 2007). The differences between those cohorts suggest that the time since diagnosis could be considered as decisive for the parental HrQoL.

According to the study's results, the most important influencing factors on the HrQoL of parents of CAH diagnosed children are the effectiveness of applied coping patterns and the intensity of needs among these parents. Regression analyses conveyed that effective coping and low intensity of particular needs are significantly correlated to an above-average HrQoL. No other influencing factors show a significant impact on the established parental HrQoL. Although the R^2 marks .404,

this regression model is adequate to sufficiently explain the influence on the parental HrQoL, since a lot of possible influencing factors were tested.

The parents in the present study valued *maintaining family integration, cooperation, and an optimistic definition of the situation* as their most helpful coping pattern. Besides, the surveyed mothers and fathers reached the highest means at the HrQoL measurement at the subdimension *satisfaction with the situation in the family*, which emphasizes the importance of well-functioning family structures. This observation is congruent with the findings of Van Schoors et al. (2019). The authors examined 115 families of children diagnosed with leukaemia or non-Hodgkin lymphoma on their experienced family functioning, perceived stress, emotional reactions, and HrQoL. They found out that mothers and fathers who experienced higher emotional closeness in their family report a better HrQoL. In particular, the perceived level of expressiveness within the family was shown to be decisive (Van Schoors et al., 2019).

Some other studies took advantage of the CHIP to investigate parental coping behaviour, e.g. McCubbin et al. (2001), Senger et al. (2016), and Clever et al. (2019). In each of these studies, the subdimension *maintaining family integration, cooperation, and an optimistic definition of the situation* was evaluated as most helpful by parents. These results underline the potential of enhancing family cohesion as a key coping pattern to improve parental HrQoL. The child's particular disease seems to be irrelevant for the parental evaluation.

Additional important coping patterns influencing the parental HrQoL are understanding the disease itself and a feeling of competence in disease management. In a cross-sectional study, Fleming et al. (2017c) examined influencing factors on the disease management ability of parents giving birth to a child with CAH. The results showed a significant, positive relationship between detailed provider instruction on managing adrenal crises and perceived parental management ability. Further, they found out that with a gain of parental management ability, the impact CAH has on the family decreases. Dellve et al. (2006) emphasized the importance of gaining competence in disease management in an interventional study. They assessed stress, well-being, and supportive resources of parents of children with a rare disability before and after going through an intensive family competence programme. After the programme, parents showed

increased knowledge about their child's disability and concerning everyday problems, more effective coping patterns, and decreased stress levels, especially among full-time working parents, fathers, and parents of particularly young children. Van Schoors et al. (2019) detected a direct connection between perceived management ability and parental HrQoL. They point out: "[…] the more one perceives the illness as uncontrollable and the less as manageable, the worse his/her quality of life" (Van Schoors et al., 2019, p. 8). These findings highlight the importance of a good understanding of the disease and competence in managing upcoming disease-related challenges for good parental HrQoL.
Concurrently, parents in the present study valued information-oriented coping as their second effective coping strategy. After family-centred coping patterns, mothers and fathers emphasized the importance of *understanding the medical situation* of their child for adapting to the new situation.
According to this, it could be suggested that parents of two children with CAH reach higher scores in this study on HrQoL scales due to pre-existing experience in taking care of a CAH affected child. However, with no significant results in regression analyses, data could not prove this thesis. Since there were only 10 families with two CAH children in the present sample, of which two families have twins and could not be included in this consideration, the analysed cohort is too small to make a representative statement. Nevertheless, it is shown that effective coping, which includes a good understanding of the disease, constitutes a crucial predictor for a good parental HrQoL.

The same accentuations can be retraced considering the parents' most urgent needs in this study. With the first three needs most frequently reported being explicit informational kind, parents made a point about receiving sufficient information around their child's disease and upcoming management challenges. The findings are comparable to the results elaborated by Wiedebusch-Quante (2009) and Schreiber (2011). They took advantage of the Bedürfnis-Skala für Eltern chronisch kranker Kinder as well. The authors surveyed parents of children diagnosed with juvenile idiopathic arthritis, diabetes mellitus, and haemophilia (Wiedebusch-Quante, 2009), respectively, a chronic kidney disease (Schreiber, 2011). In both studies, the parents' most intense needs are concurrent with the requirements revealed at the present examinations. This suggests that regardless of a specific

disease, parents of chronically ill children deal with similar questions and struggle with similar problems. As elaborated earlier, well-informed parents state a higher perceived management ability and consequently reach higher HrQoL scores (Fleming et al., 2017a, Van Schoors et al., 2019). Therefore, it is crucial to convey thorough and understandable information and, in this way, meet parental needs as early as possible.

In the context of information collection, parents in the present study accentuate contact with other affected families. Almost one third stated to get into active contact with other parents in the same situation through self-help groups. In free-text fields, the surveyed mothers and fathers often mentioned the acknowledged and registered support group: AGS- Eltern- und Patienteninitiative e.V. (https://www.ags-initiative.de) - a union of CAH patients and parents to build a forum for the exchange of information and to connect affected families by organizing meetings in Germany, Austria, and Switzerland. Further, this association also created a detailed information brochure about CAH. Parents described the AGS-Eltern- und Patienteninitiative e.V. as very helpful to gain information, practical advice, and in addition, social support. However, around half of the parents (53.3%) did not contact other affected families. According to Kang et al. (2012), parent-to-parent communication is a meaningful intervention to improve knowledge, self-efficacy, and quality of life of mothers of ill children. Kang et al. (2012) examined mothers of young haemophilia patients before and after an intervention programme compared to a control group receiving only education materials. The intervention programme included guided self-help group sessions twice a week for over five weeks. Other studies point out the positive effect of self-help groups on the perceived HrQoL of patients themselves (Kang et al., 2005, Chaveepojnkamjorn et al., 2009). All these findings suggest a recommendation of parent-to-parent contact for mothers and fathers of CAH diagnosed children being desirable as well.

Contrary to expectations, parents in this study did not declare a high need for psychosocial support at the survey time. Altogether, only 15.7% of the mothers and fathers used psychosocial consultation in the past. However, about two thirds of this 15.7% evaluate it as helpful or very helpful. Studies showed that psychological support, e.g. through cognitive behavioural therapy, is highly recommended for parents of children with chronic health issues (Wong and Poon, 2010, Levy et al., 2017). Parents of children with CAH are at risk of developing psycho-pathological

symptoms like depression, anxiety or posttraumatic stress symptoms, especially when they receive the final diagnosis disclosure (Barg et al., 2010, Suorsa et al., 2015). Therefore, psychosocial support seems to be prophylactically advisable from the start. However, it can be assumed that the parents' need for psychosocial help was no longer present at the time of the survey since, in most cases, many years have passed after the disclosure of the child's diagnosis. Another reason could be that there was an impeded access to psychosocial consultation. Only 12.7% of the parents in the present study received an offer of psychosocial support at the ward at their first clinical stay. Consequently, early offering of psychosocial support could be a topic with scope for improvement.

In conclusion, the study's results show that both, effective coping patterns as well as rapid fulfilment of parental needs are decisive for good parental HrQoL. These findings are consistent with the current state of research on this topic. However, time seems to be an additional important influencing factor. It takes time to find individually fitting coping patterns and develop disease management ability through an active learning process. At the end of the survey paper, parents in this study had the possibility to add comments. An often-mentioned comment was that after receiving the child's presumptive diagnosis, it was challenging for the mothers and fathers to get along with the situation. Though they emphasized that at the time of the survey they were doing very well and they do not anymore experience extensive daily life restrictions through their child's disease. One parent of a child with CAH from the studies of Fleming et al. (2017a, p. 6) summarized: " 'In the first few months [after diagnosis] it was trying to understand it. It was educating. It was learning about it. It was researching. It was trying to understand how it happened, why it happened ... The first couple of years were the roughest but as he's grown and gotten better, it is easier' ".

Similar observations can be retraced in various publications. Regardless of a particular disease, the disclosure of a child's diagnosis often constitutes a shocking event for parents (Fonseca et al., 2013, Lundberg et al., 2017, Goldbeck et al., 2001, Talakoub and Nasiri, 2012, Caplan, 2013). Especially at the beginning of their child's disease, parents undergo many different feelings like uncertainty, helplessness, and insecurity. Many questions around the disease itself, the disease management, and possibilities to cope with the new situation are yet unresolved (Lundberg et al., 2017,

Van Schoors et al., 2019, Suorsa et al., 2015). A situation like this produces high parental stress and intense adverse emotional reactions like anxiety, sadness, despair, and frustration in affected families (Fonseca et al., 2013, Dellve et al., 2006, Talakoub and Nasiri, 2012). Studies showed that parents going through times of great insecurities and helplessness have a reduced HrQoL (Van Schoors et al., 2019, Knibb et al., 2016). However, with passing time, the parental HrQoL mainly increases again as parents get familiar with the new situation and learn to manage associated obstacles. This process can be observed in parents being confronted with common chronic diseases of their child (Goldbeck et al., 2001, Van Schoors et al., 2019). Further, it can be assumed for parents of children being diagnosed with CAH (Witt et al., 2018, Fleming et al., 2017a, Ellens et al., 2017). According to my best state of knowledge, no longitudinal studies examining this specific subject exist at this point. However, Fleming et al. (2017a, p. 9) comment their findings: "[…] over time parents adapt to the management challenges of having a child with CAH […]".

It can be assumed that the parents in the present study had already overcome the critical phase of managing the upcoming challenges around the time of disclosure of the disease. They showed good adaptions to their new life circumstances through effective coping patterns and an only as average measured intensity of needs. Passing time seems to be an essential factor influencing parental HrQoL. However, further research should be initiated to confirm these considerations and investigate parental well-being using a prospective study design.

5.2 Practical Recommendations

Regarding the critical phase around the time of the disclosure of the diagnosis parents in this study expressed their needs in free-text fields at the end of the present survey very clearly. They demand understandable information, well-trained physicians, psychosocial support, and a rapid referral to a specialist like a paediatric endocrinologist. To develop and suggest demand-responsive, purposeful interventions for parents of children with a recent CAH diagnosis, it is helpful to consider the current state of the provision of care for these families.

Current State of the Art:

According to the present study, parents of children with CAH mostly found out about the possible diagnosis of their child through physicians at their maternity clinic after a conspicuous result in new-born screening. The first disclosure happened at a median of 6.5 days after the birth of the child. The bearer of the final CAH diagnosis in most cases was an assistant medical director or head doctor at a paediatric clinic. These findings show that the maternity and paediatric clinic, both play an important role in providing first care for affected families.

The results show that most of the parents felt sufficiently informed after disclosing the final diagnosis of their child. Nevertheless, an alarming proportion of 23.5% of mothers and fathers state a lack of adequate information. Consequently, improving the mediation of first information depict an important part for improving the provision of care for these families. All-embracing information is crucial to avoid dangerous consequences regarding the health of CAH children, which underlines the importance of this aspect of care. Even after a mean time of 11 years after the disclosure of the CAH diagnosis of their child, mothers and fathers still state more information concerning the disease being their primary need. This implies that with a complex disease like CAH, new arrival questions and informational needs are shared continuously. With nearly 70% of the parents naming a physician as their main source of information, it seems important for parents to have a constant competent medical contact for guiding through the obstacles of CAH over time.

After conducting the final CAH diagnosis at the paediatric clinic, most families were referred to an outpatient paediatric endocrinologist. In the present study, the mean time until the first consultation shows a wide variation, with a median of 40 days from birth. Fast contact with an experienced paediatric endocrinologist is yet very important since CAH constitutes a rare chronic health condition. Physicians, especially at smaller clinics, often lack the knowledge and experience in treating a such rare and complex disease (Lundberg et al., 2017, Buchbinder and Timmermans, 2012). However, well-founded knowledge seems to be especially important before informing the parents about their child's presumptive diagnosis to meet all upcoming needs adequately.

It can be considered that with a rare disease like CAH, all-embracing and experience-based information can be especially given through an expert, e.g. the outpatient paediatric endocrinologist. With a median time of 40 days until meeting

the outpatient paediatric endocrinologist, mothers and fathers in the present study had to endure a long time on their own until their informational needs could be fully met. Additionally, psychosocial support or possible interventions to improve parental coping patterns were barely offered, as mentioned before. Therefore, it is desirable to provide first information material and further details on self-help groups etc., parallel to conducting the final diagnosis to bridge the time until meeting the outpatient paediatric endocrinologist.

All of this illustrates the current gap between recommendable theoretical findings and practical action in the care of CAH affected families. Therefore, suggestions for improved care based on scientific data are listed in the following.

<u>Recommendations for Improving the Provision of Care for CAH Affected Families:</u>

- Rapid consultation of specialists (paediatric endocrinologist)
- Offering a constant medical contact for parents, ideally a paediatric endocrinologist
- Recurrent sensitization of physicians at maternity & paediatric clinics for:
    - the process of new-born screening and knowledge on detectable diseases like CAH - cf. (Nennstiel et al., 2020)
    - the possibility to consult experts through the whole diagnosis process
    - sensitively conducting a (presumptive) diagnosis
    - addresses to pass on to affected families for further treatment
- Early offer of psychosocial support for the entire family at times of presumptive diagnosis
- Improved way of communicated information: timely, understandable, thorough, without too many specialist terms, directly at conducting the final diagnosis
- Deliver written material with accessible and understandable information about the disease, upcoming obstacles with suggested solutions, practical advice, directly at conducting the final diagnosis
- Imparting recommendations for further sources for verified, science-based, understandable research - e.g. internet pages, books, brochures
- Early encouragement to participate in self-help groups, directly at conducting the final diagnosis - e.g. AGS-Eltern- und Patienteninitiative e.V.

- Imparting phone numbers to consult / addresses for places to go in case of emergency
- Early interventions to tighten family solidarity and to improve communication within the family (family-based coping)
- Early and repeated training for the management of adrenal crises (information-based coping)
- Offering a structured, short information brochure with information about CAH and instructions in cases of emergency for handing over to (nursery) school, and other supervisory staff

5.3 Strengths and Limitations

To the best of my knowledge, this is the first study taking a closer look at the HrQoL of parents of CAH diagnosed children. The findings provide important impulses for improving the situation of CAH affected families since decisive influencing factors on the parental HrQoL were found.

Even though the examinations concern a rare disease, many participants could be gathered for this study. Questionnaire packages were addressed to 83.9% of all registered parents with a child diagnosed with CAH through new-born screening in Bavaria. With a response rate of 43.8%, the sample covers a large part of affected families in this region. Additionally, with three out of 105 questionnaires, the exclusion rate was very low. Further, contrary to many other studies, both, mothers and fathers were included in the survey. The questionnaire package consisted of a HrQoL measurement and many more important aspects as well, like sociodemographic and clinical assessments, questions about circumstances regarding the diagnosis process, used parental coping patterns, and the intensity of particular needs of the parents. This approach opens up the possibility to make well-founded statements about the psychosocial situation of parents with CAH diagnosed children. Another strength of this study is grounded in applied methods. The use of a Bonferroni-adjustment for multiple testing ensures the prevention of possible interference factors to a large extent.

Nevertheless, various limitations to the study have to be acknowledged. Since the study format was retrospective and not based on objective data, a certain recall bias cannot be excluded. Further, even though there was a satisfying number of returns, it is possible that the parents who did not participate maybe struggle with their

situation the most and did not have capacity for answering the survey. An important fact to add is that the childrens' age in this study spread on a wide range (.7-19.4 years). Therefore, parents being newly confronted with the diagnosis of their child and parents who had years to adapt to the new situation both filled in the questionnaires, what makes the study population heterogeneous. Apart from this, the present study describes a regional sample, including only parents living in Bavaria, Germany.

Yet, this study still delivers a profound gain of knowledge about the psychosocial situation of CAH affected families and elaborates important impulses to improve the provision of care. Further studies should concentrate on accompanying families with CAH diagnosed children early on and in other federal states and countries. A longitudinal format is suggestable.

6. Conclusion

To provide adequate and all-embracing medical care, it is crucial to contemplate the clinical values and the psychosocial situation of a patient (WHO, 2006, Bullinger and Quitmann, 2014). If a child gets the diagnosis of a chronic disease, it often marks an unexpected, extraordinary and stressful change in the prevailing family life system. Several studies showed that parents of children diagnosed with a chronic health condition are at risk of experiencing significant deteriorations in their physical, emotional, and social well-being (Cousino and Hazen, 2013, Grootenhuis and Bronner, 2009). Since CAH constitutes a rare chronic disease with a potentially life-threatening character, parents must overcome additional obstacles in caring for their child. Thus, they are even more at risk of undergoing psychological strain and a decline in their HrQoL (Dellve et al., 2006, Jaffe et al., 2010, de Silva et al., 2014). Parental well-being is a fundamental basis for a child's psychosocial development (Grootenhuis and Bronner, 2009, Epifanio et al., 2015). Therefore, it is crucial to consider the psychosocial situation of parents of children with a CAH diagnosis.

The present study is the first to extensively examine the HrQoL of parents giving birth to a child diagnosed with CAH. While taking a closer look to the HrQoL, coping patterns, and particular needs of these parents, this study provides essential impulses to improve the provision of care for affected families.

The results show that despite the complex and seemingly threatening nature of CAH, parents are doing pretty well and do not experience extensive restrictions in their daily lives through their child's disease. Some indications are showing that parents of a child with CAH experience difficulties in dealing with the situation around the time of diagnosis (Barg et al., 2010, Boyse et al., 2014, Witt et al., 2018). The present data though implicate an above-average HrQoL in these mothers and fathers, surveyed on average 11 years after the initial diagnosis.

Possible causes can be seen as two-fold. On the one hand, the established parental HrQoL is strongly influenced by the effectiveness of the mother’s and father’s applied coping patterns. Parents in the present study described their coping mechanisms as helpful so that an effective coping can be assumed in them.

On the other hand, the intensity of needs in parents of a child with CAH depict a significantly influencing factor on their perceived HrQoL. The only as average evaluated intensity of needs in this study contributes to the good parental HrQoL. Hence, this study verifies the importance of helpful coping patterns and the rapid fulfilment of parental needs for maintaining a good and stable HrQoL of parents with a child diagnosed with CAH.

Therefore, it is crucial to develop demand-responsive interventions to support affected families from the very beginning and early counteract a decrease of the parental HrQoL. According to the present findings, suggestions to intervene should especially include adequate interventions concentrating on family- and information-based coping. Early intervention programs should focus on tightening family solidarity, improving open communication within the family, and providing early training to reinforce parental disease management ability. Further studies should concentrate on the development and effectiveness of such interventions.

7. Summary

- ENGLISH

The endocrine disorder CAH signifies a rare chronic disease with a worldwide incidence between 1:14000 to 1:18000 births (Speiser et al., 2018). Affected female new-borns can show ambiguous external genitalia and in both sexes with the salt-wasting form, hazardous adrenal crises developed postnatally (Stokowski, 2009). The diagnosis of CAH constitutes a profound change in parental thinking. The expectant mothers and fathers must deal not only with the new task of becoming a parent but also with unexpected additional responsibilities. Studies show an increasing strain in these parents, as well as health-related distress and a high need of information and psychosocial support (Grootenhuis and Bronner, 2009, Witt et al., 2018, Boyse et al., 2014). With the new-born screening there exist a possibility to detect CAH early and thus reduce grave consequences (Odenwald et al., 2015).

This doctoral thesis focuses on the quantitative part of a retrospective cross-sectional study examining the HrQoL, coping, and needs of parents caring for a child with CAH diagnosed by new-born screening in Bavaria. Between 09/2018 and 09/2019 102 parents of 70 children diagnosed with CAH answered specific questionnaires. The age of the respective children ranged from a few months to 19 years. 81% of the 32 girls and 38 boys had further a salt-wasting syndrome. Besides, 72% of the girls were born with virilized external genitalia.

The results show that despite the complex and seemingly threatening nature of CAH, parents are doing pretty well and do not experience extensive restrictions in their daily lives through their child's disease. Compared to parents of a child diagnosed with an oncological disease, diabetes mellitus, or epilepsy (cf. Goldbeck and Storck, 2002), mothers and fathers in this study reached significantly higher HrQoL scores. Decisive for the above-average parental HrQoL were an effective coping behaviour and an only as average evaluated intensity of needs of the parents.

These findings verify the importance of helpful coping patterns and a rapid fulfilment of parental needs for maintaining a good and stable HrQoL of parents with a child diagnosed with CAH. The parental psychosocial situation plays an important part in the future coping abilities of the child (Grootenhuis and Bronner, 2009). Therefore, it is crucial to strengthen the parental HrQoL to build a reasonable basis for a healthy upbringing and improve the medical care of CAH diagnosed children. Future studies should focus on the development of interventions to support affected families from the very beginning.

- DEUTSCH

Das Adrenogenitale Syndrom mit 21-Hydroxylase-Defekt [AGS] beschreibt eine seltene chronische Stoffwechselerkrankung, die weltweit mit einer Inzidenz von circa 1:14000 bis 1:18000 Geburten auftritt (Speiser et al., 2018). Betroffene Mädchen können mit einem mehrdeutigen äußeren Genital bei der Geburt auffallen. Des Weiteren können Kinder beider biologischer Geschlechter postnatal von verheerenden adrenalen Krisen betroffen sein (Stokowski, 2009). Die Diagnose AGS stellt einen drastischen Umschwung im elterlichen Denken dar. Die werdenden Mütter und Väter sehen sich nicht nur mit der neuen Aufgabe der Elternschaft, sondern zusätzlich mit unerwarteten weiteren Herausforderungen konfrontiert. Nicht selten geht dies mit krankheitsbezogenem Stress und einem großen Informations- und Unterstützungsbedarf der Eltern einher (Grootenhuis and Bronner, 2009, Witt et al., 2018, Boyse et al., 2014). Mit dem Neugeborenenscreening können betroffene Kinder früh entdeckt und schwerwiegende Folgen der Erkrankung reduziert werden (Odenwald et al., 2015).

Diese Doktorarbeit behandelt den quantitativen Anteil einer retrospektiven Querschnittstudie, die die gesundheitsbezogene Lebensqualität [HrQoL], Bewältigungsmechanismen und Bedürfnisse der Eltern eines Kindes mit AGS, das im Neugeborenenscreening in Bayern diagnostiziert wurde, untersucht. In dem Zeitraum 09/2018 bis 09/2019 haben 102 Eltern von 70 Kindern mit AGS spezifische Fragebögen beantwortet. Das Alter der Kinder in den untersuchten Familien erstreckte sich hierbei auf eine Spanne zwischen wenigen Monaten und 19 Jahren. Bei 81% der insgesamt 32 Mädchen und 38 Jungen wurde zusätzlich ein Salzverlust-Syndrom festgestellt. Außerdem wurden 72% der Mädchen mit einem vermännlichten Genital geboren.

Die Ergebnisse zeigen, dass die befragten Eltern trotz des bedrohlichen Charakters eines AGS durch die Erkrankung ihres Kindes keine weitreichenden Einschränkungen ihres Alltags erleben. Verglichen mit Eltern eines Kindes mit einer onkologischen Erkrankung, Diabetes mellitus oder Epilepsie (vgl. Goldbeck and Storck, 2002) erreichten die Mütter und Väter in der vorliegenden Studie signifikant höhere HrQoL-Werte. Maßgeblich für das Erreichen der überdurchschnittlichen HrQoL waren wirkungsvolle Bewältigungsstrategien und eine lediglich als mittelmäßig evaluierte Intensität spezifischer Bedürfnisse der Eltern.

Dies betont die Bedeutung von wirkungsvollen Bewältigungsstrategien und dem schnellen Eingehen auf spezifische Bedürfnisse für eine gute HrQoL von Eltern eines Kindes mit AGS. Die elterliche psychosoziale Situation steht in engem Zusammenhang mit der kindlichen Bewältigung der eigenen Erkrankung (Grootenhuis and Bronner, 2009). Demnach ist die Beachtung dieser Faktoren maßgeblich, um die HrQoL der Eltern zu fördern und somit in einem ganzheitlichen Versorgungskonzept das gesunde Aufwachsen des Kindes zu gewährleisten. Zukünftige Studien sollten sich mit der Entwicklung von Interventionen zur frühzeitigen Unterstützung betroffener Familien befassen.

8. List of Abbreviations

21-OH	21-hydroxylase enzyme
AGS	Adrenogenitales Syndrom mit 21-Hydroxylase-Defekt
AI	adrenal insufficiency
BAI	The Beck Anxiety Inventory
BDI-II	Beck Depression Inventory – 2nd Edition
CAH	congenital adrenal hyperplasia with 21-hydroxylase deficiency
CBAQ	Child Behaviour and Attitudes Questionnaire
CHIP	Coping Health Inventory for Parents
CI	confidence interval
CoR	coefficient of regression
df	degrees of freedom
DRS	Decisional Regret Scale
DSD	disorder of sex development
EBI	Eltern-Belastungs-Inventar
FAD	Family Assessment Device
FLZ	Fragebogen zur Lebenszufriedenheit
GSES	Generalized Self-Efficacy Scale
HrQoL	health-related quality of life
IES-R	Impact of Event Scale Revised
LGL	Bayerisches Landesamt für Gesundheit und Lebensmittelsicherheit, Screeningzentrum Bayern
M	arithmetic mean value
max	maximum value
min	minimum value
NaCl	sodium chloride
OSS-3	Oslo 3-items Social Support Scale
P	p-value
PPUS	Parental Perceptions of Uncertainty Scale
QoL	quality of life
SD	standard deviation
SE	standard error
SF-36	Short Form health survey (36items)
t	t-value
UKE	University of Hamburg-Eppendorf
ULQIE	Ulmer Lebensqualitätsinventar für Eltern chronisch kranker Kinder
WHO	World Health Organization

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11. Attachment

11.1 Questionnaire Package

Wie geht es Eltern von Kindern mit einem im Neugeborenenscreening diagnostizierten Adrenogenitalen Syndrom (AGS)?

Fragebogen für Elternteil 1

Fragebogen-Nr.* .....

* Diese Nummer wird ausschließlich verwendet, um anonym abzugleichen, welche Fragebögen gemeinsame Kinder betreffen

Klinische & Wissenschaftliche Projektleitung
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Heutiges Datum: .....

Wer füllt den Fragebogen aus?

- ○ Mutter
- ○ Vater

Universitätsklinikum Erlangen | Universitätsklinikum Hamburg-Eppendorf | Bayerisches Landesamt für Gesundheit und Lebensmittelsicherheit

Liebe Eltern!

Wie geht es Ihnen? Wie fühlen Sie sich?

Das möchten wir von Ihnen wissen. Deshalb möchten wir Sie herzlich dazu einladen, unseren Fragebogen auszufüllen. Bei den Fragen geht es um Ihr Leben im Allgemeinen, Ihre Stimmungslage und den Umgang mit der Krankheit Ihres Kindes.
Aufgrund Ihrer Antworten möchten wir einschätzen, wie es Ihnen mit der Diagnose Ihres Kindes geht und gezielt Unterstützungsangebote für betroffene Eltern entwickeln.

Für jeden leiblichen Elternteil gibt es einen eigenen Fragebogen; bitte bearbeiten Sie diese getrennt voneinander, da uns die Sichtweise von Vätern und Müttern interessiert.

Was ist zu tun? Füllen Sie bitte den beigefügten Fragebogen aus und schicken Sie diesen im vorfrankierten Rückumschlag an das Universitätsklinikum Hamburg-Eppendorf.

Sollten Sie beim Ausfüllen des Fragebogens den Bedarf an psychosozialer/psychologischer Unterstützung verspüren, kontaktieren Sie gerne Frau Dr. Julia Quitmann als zentrale Ansprechpartnerin (Tel. 040 7410 52789, E-Mail: j.quitmann@uke.de).
Bei weiteren Fragen oder Unklarheiten können Sie sich auch an das Screeningzentrum wenden (Tel. 09131 6808 5188, E-Mail: birgit.odenwald@lgl.bayern.de).

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Mit freundlichen Grüßen

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Universitätsklinikum Hamburg-Eppendorf

Hinweise zum Ausfüllen

- ✓ Gehen Sie bitte den Fragebogen Frage für Frage durch.
- ✓ Die Fragen können Sie durch Ankreuzen beantworten. Bitte kreuzen Sie zu jeder Frage nur eine Antwort an.
- ✓ Es gibt keine richtigen oder falschen Antworten. Es gibt nur Ihre Meinung und die ist für uns wichtig.
- ✓ Antworten Sie bitte offen und ehrlich; so, wie es für Sie tatsächlich gilt. Alle Ihre Angaben werden streng vertraulich behandelt und die Datenverarbeitung erfolgt vollkommen anonym.
- ✓ Vielleicht passen einige Fragen nicht besonders gut auf Sie bzw. Ihr Kind. Kreuzen Sie aber trotzdem immer eine Antwort an, und zwar die, welche noch am ehesten auf Sie bzw. Ihr Kind zutrifft.
- ✓ Bitte versuchen Sie diesen Fragebogen vollständig auszufüllen.

Allgemeine Fragen

A1	Ich bin…	○ männlich ○ weiblich
A2	Ich bin …	…….. Jahre alt
A3	Ich habe …	..…… Kinder (Anzahl)
A4	Ich bin ...	○ verheiratet ○ geschieden ○ verwitwet ○ ledig
A5	Ich lebe ständig mit einem Partner oder einer Partnerin zusammen	○ ja ○ nein
A6	Ich lebe in einem gemeinsamen Haushalt mit … … dem Kind, das AGS hat. … dem anderen leiblichen Elternteil dieses Kindes	 ○ ja ○ nein ○ ja ○ nein
A7	Ich bin geboren …	○ in Deutschland ○ in einem anderen europäischen Land ○ außerhalb von Europa
A8	Der Wohnort, in dem ich meinen Lebensmittelpunkt habe …	○ Großstadt (≥ 100.000 Einwohner) ○ Mittelstadt (≥ 20.000 bis unter 100.000 Einwohner) ○ Kleinstadt (≥ 5.000 bis unter 20.000 Einwohner) ○ Land (unter 5.000 Einwohner)
A9	Ich (Elternteil) *Wenn Sie an einer Erkrankung leiden, um welche Erkrankung handelt es sich?* ……..…….....	○ bin gesund ○ habe eine chronische Erkrankung
A10	Mein höchster Bildungsabschluss	○ kein Abschluss ○ Hauptschulabschluss ○ mittlere Reife ○ Abitur ○ Studienabschluss ○ anderer; …….
A11	In den letzten 12 Monaten war ich / habe ich …	○ in Vollzeit gearbeitet ○ in Teilzeit gearbeitet ○ in Elternzeit ○ krank ○ Student/in ○ arbeitssuchend/- los ○ sonstiges; ……

A12	Haushaltsnettoeinkommen pro Monat	○ < 1.300 € ○ 1.300 bis < 2.000 € ○ 2.000 bis < 4.500 € ○ 4.500 € und mehr
A13	Erhalten Sie staatliche oder andere finanzielle Untersetzung? *Wenn ja, welche finanzielle Unterstützung erhält Ihre Familie?* ……	○ nein ○ ja
A14	Haben Sie schon einmal psychosoziale Beratungsangebote in Anspruch genommen? (z.B. sozialpädagogisch oder psychologisch)	○ ja (*wenn ja, bitte weiter mit Frage A14.1*) ○ nein (*wenn nein, bitte weiter mit Frage A15*)
	Wenn ja:	
A14.1	Welche Art von Beratung haben Sie in Anspruch genommen?	○ psychologische Beratung/Therapie ○ Gruppengespräche ○ Einzelgespräche ○ Familienberatung ○ Sonstige; ……
A14.2	War die Beratung/Therapie für Sie hilfreich und hat Ihnen weitergeholfen?	○ gar nicht ○ wenig ○ teilweise ○ viel ○ sehr viel
A15	Welche Quellen nutzen Sie, um sich über die Erkrankung Ihres Kindes zu informieren?	○ Internet ○ Informationsmaterial von Arzt, Ärztin oder Klinik ○ Fachbücher ○ keine ○ sonstige;……
A16	Wie tauschen Sie sich mit anderen Eltern von Kindern mit AGS aus?	○ Selbsthilfegruppe ○ Internet-Foren ○ Soziale Netzwerke ○ gar nicht ○ sonstiges; ……

Allgemeine Fragen zu Ihrem ersten (älteren) Kind mit AGS *

B1	Wann ist Ihr Kind geboren?	……. / …….. (*Monat / Jahr*)
B2	Das Kind ist	O ein Mädchen O ein Junge
B3	War die Geburt … … eine Mehrlingsgeburt? … eine Frühgeburt? (vor der 37. vollendeten Schwangerschaftswoche) … ein Kaiserschnitt?	O nein O ja O nein O ja O nein O ja
B4	Gab es nach der Geburt Komplikationen?	O nein, es gab keine Komplikationen O ja, Komplikationen bei der Mutter O ja, Komplikationen beim Kind *Wenn ja, welche?* ……..….. ……..
B5	Wann haben Sie erfahren, dass Ihr Kind möglicherweise ein AGS hat?	O noch in der Klinik O Zuhause O anderes;……
B6	Wie alt war das Kind, als man Ihnen die Verdachtsdiagnose AGS mitgeteilt hat?	…… *(Alter in Tagen)*
B7	Wie haben Sie die Verdachtsdiagnose erfahren?	O am Telefon O im direkten Gespräch O anderes;……
B8	Wer hat Ihnen die Verdachtsdiagnose mitgeteilt?	O Hebamme oder Entbindungspfleger O Krankenschwester oder -pfleger O Ärztin oder Arzt der Geburtsklinik O Kinderärztin oder Kinderarzt O andere; ……
B9	Wurde Ihr Kind für weitere Untersuchungen zur Sicherung der Diagnose in einer Klinik stationär aufgenommen?	O ja (*wenn ja, bitte weiter mit Frage B10)* O nein *(wenn nein, bitte weiter mit Frage B15)*
B10	*Wenn ja:* Wie alt war Ihr Kind bei der Aufnahme in der Klinik?	…… *(Alter in Tagen)*
B11	In welcher Art von Klinik wurde Ihr Kind aufgenommen?	O Spezialisiertes Zentrum für Kinder-Endokrinologie O Kinderklinik O andere; ……
B12	Wurden Sie zusammen mit dem Kind stationär aufgenommen?	O ja O nein
B13	Gab es während des Klinikaufenthaltes Komplikationen?	O ja O nein *Wenn ja, welche?* ……..….. ……..

* Wenn Sie sich untereinander absprechen, müssen die Fragen zum Kind (B1-B26) nur von einem Elternteil beantwortet werden.

B14	Haben Sie auf der Station zusätzlich zur medizinischen Versorgung weitere Hilfe bekommen von …?	O einem Psychologen oder einer Psychologin O einem Sozialpädagogen oder einer Sozialpädagogin O anderen; …… O Ich habe keine weitere Hilfe bekommen
B15	Welche Medikamente hat Ihr Kind bekommen, nachdem die Diagnose AGS gesichert war?	O Hydrocortison O Fludrocortison (Astonin H) O Kochsalzlösung O andere; ……
B16	Wer hat Ihnen die endgültige Diagnose AGS erstmals mitgeteilt?	O Ärztin/Arzt für Kinderendokrinologie O Stationsärztin/-arzt O Oberärztin/-arzt oder Chefärztin/-arzt O Stationsschwester/-pfleger O andere; ……
B17	Wurden Sie ausreichend über die Erkrankung informiert?	O ja O nein
B18	Hat man Ihnen gesagt, dass die Krankheit vererbt wird?	O ja O nein
B19	Haben Sie die Diagnose Ihren Eltern / Verwandten sofort mitgeteilt?	O ja O nein
B20	Wie alt war Ihr Kind, als Sie nach der Entlassung das erste Mal ambulant bei einem Arzt oder einer Ärztin für Kinderendokrinologie waren?	…… *(Alter in Tagen)*
B21	Musste Ihr Kind im ersten Lebensjahr außer zur Sicherung der Diagnose stationär aufgenommen werden? *Wenn ja, warum?* ……	O ja O nein
B22	War Ihr Kind nach dem ersten Lebensjahr wegen einer Salzverlustkrise oder Unterzuckerung im Krankenhaus?	O ja, einmal O ja, mehrmals O nein
B23	Hat Ihr Kind leibliche Geschwister? *Wenn ja, wie ist die Position dieses Kindes in der Geschwisterfolge?*	O ja O nein *(Nummer 1, 2, 3 etc.)* ……
B24	Gibt es weitere Familienmitglieder mit der Diagnose AGS? *Wenn ja, wer ist noch betroffen?*	O ja O nein ……
	Die folgenden Fragen betreffen nur Mädchen mit AGS	
B25	War das äußere Genital vermännlicht?	O ja O nein
B26	Wurde schon eine Operation des Genitales durchgeführt? *Wenn ja, Alter bei der Operation?*	O ja O nein ……

Allgemeine Fragen zu Ihrem zweiten (jüngeren) Kind mit AGS *

C1	Wann ist Ihr Kind geboren?	……. / …….. (*Monat / Jahr*)
C2	Das Kind ist	O ein Mädchen O ein Junge
C3	War die Geburt … … eine Mehrlingsgeburt? … eine Frühgeburt? (vor der 37. vollendeten Schwangerschaftswoche) … ein Kaiserschnitt?	O nein O ja O nein O ja O nein O ja
C4	Gab es nach der Geburt Komplikationen?	O nein, es gab keine Komplikationen O ja, Komplikationen bei der Mutter O ja, Komplikationen beim Kind *Wenn ja, welche?* …… ……
C5	Gab es während des Klinik-aufenthaltes Komplikationen?	O ja O nein *Wenn ja, welche?* …… ……
C6	Welche Medikamente hat Ihr Kind bekommen, nachdem die Diagnose AGS gesichert war?	O Hydrocortison O Fludrocortison (Astonin H) O Kochsalzlösung O andere; ……
C7	Musste Ihr Kind im ersten Lebensjahr außer zur Sicherung der Diagnose stationär aufgenommen werden?	O ja O nein *Wenn ja, warum?* …… ……
C8	War Ihr Kind nach dem ersten Lebens-jahr wegen einer Salzverlustkrise oder Unterzuckerung im Krankenhaus?	O ja, einmal O ja, mehrmals O nein
	Wie ist die Position dieses Kindes in der Geschwisterfolge?	(Nummer 1, 2, 3 etc.) ……
	Die folgenden Fragen betreffen nur Mädchen mit AGS	
C9	War das äußere Genital vermännlicht?	O ja O nein
C10	Wurde schon eine Operation des Genitales durchgeführt?	O ja O nein
	Wenn ja, Alter bei der Operation?	……

* Wenn Sie sich untereinander absprechen, müssen die Fragen zum Kind (C1-C10) nur von einem Elternteil beantwortet werden.

Über Ihr Lebensgefühl / Ihre Stimmung

*Die folgenden Fragen beziehen sich auf Ihr Befinden **in den letzten 7 Tagen.** Bitte beantworten Sie die Fragen so, wie es Ihrem eigenen Empfinden am besten entspricht.*

	In der letzten Woche …	nie	selten	manchmal	oft	immer
D1	… war ich aktiv und voller Energie	O	O	O	O	O
D2	… hatte ich körperliche Beschwerden	O	O	O	O	O
D3	… fühlte ich mich erschöpft	O	O	O	O	O
D4	... konnte ich gut schlafen	O	O	O	O	O
D5	… konnte ich gut essen	O	O	O	O	O
D6	… habe ich mich fit gefühlt	O	O	O	O	O
D7	… hatte ich Schmerzen	O	O	O	O	O
D8	… konnte ich mich gut konzentrieren	O	O	O	O	O
D9	… habe ich mir Sorgen gemacht	O	O	O	O	O
D10	… hatte ich Mühe, mich zu etwas aufzuraffen	O	O	O	O	O
D11	… war ich hoffnungsvoll und zuversichtlich	O	O	O	O	O
D12	… war ich belastbar	O	O	O	O	O
D13	… war ich reizbar und nervös	O	O	O	O	O
D14	… konnte ich eigene Wünsche und Bedürfnisse verwirklichen	O	O	O	O	O
D15	… war ich niedergeschlagen und unglücklich	O	O	O	O	O
D16	… hatte ich genügend Gelegenheit, Freunde und Bekannte zu treffen	O	O	O	O	O

	In der letzten Woche …	nie	selten	manchmal	oft	immer
D17	… habe ich mich im Kreis meiner Familie wohlgefühlt	O	O	O	O	O
D18	… war mein Verhältnis zu meinem Kind / meinen Kindern zufriedenstellend	O	O	O	O	O
D19	… war ich mit meiner Partnerschaft zufrieden	O	O	O	O	O
D20	… hatte ich jemanden, mit dem ich vertrauensvoll reden konnte	O	O	O	O	O
D21	… geriet ich schnell in Streit und Auseinandersetzungen	O	O	O	O	O
D22	… war ich im Beruf / im Haushalt voll leistungsfähig	O	O	O	O	O
D23	… konnte ich die wichtigsten anstehenden Aufgaben erledigen	O	O	O	O	O
D24	… hat mich die Betreuung meines kranken Kindes stark belastet	O	O	O	O	O
D25	… hatte ich ausreichend freie Zeit für mich persönlich	O	O	O	O	O
D26	… hatte ich genügend Zeit für meinen Partner / meine Partnerin	O	O	O	O	O
D27	… fühlte ich mich durch die Krankheit meines Kindes belastet	O	O	O	O	O
D28	… konnten wir uns in der Familie gegenseitig unterstützen	O	O	O	O	O
D29	… konnten wir in der Familie offen miteinander reden	O	O	O	O	O

Fragen zum Umgang mit der Diagnose

Bitte lesen Sie die unterstehende Liste von Bewältigungsmöglichkeiten, d.h. wie Sie oder die gesamte Familie mit den Anforderungen im Zusammenhang mit dem AGS Ihres Kindes umgeht.
Bitte geben Sie für jede Bewältigungsweise, die Sie verwendet haben oder derzeit verwenden, an, wie hilfreich sie für Sie war bzw. ist.

	Wie hilfreich war oder ist für Sie …	gar nicht hilfreich	kaum hilfreich	etwas hilfreich	sehr hilfreich	habe ich nicht ausprobiert	war nicht möglich
E1	die Stabilität der Familie aufrechterhalten	O	O	O	O	O	O
E2	Beziehungen und Freundschaften pflegen, die mir das Gefühl geben, wichtig und geachtet zu sein	O	O	O	O	O	O
E3	Meinem Partner / meiner Partnerin vertrauen, dass er/sie mich und mein Kind oder meine Kinder unterstützt	O	O	O	O	O	O
E4	schlafen	O	O	O	O	O	O
E5	beim Klinikbesuch mit dem ärztlichen, pflegerischen oder psychologischen Personal sprechen	O	O	O	O	O	O
E6	daran glauben, dass es meinem Kind bald besser geht	O	O	O	O	O	O
E7	berufstätig sein	O	O	O	O	O	O
E8	Stärke zeigen	O	O	O	O	O	O
E9	Geschenke für mich oder die anderen Familienmitglieder kaufen	O	O	O	O	O	O
E10	mit anderen Personen in ähnlicher Lage sprechen	O	O	O	O	O	O
E11	die Medikamente meines Kindes in Ordnung halten	O	O	O	O	O	O
E12	essen	O	O	O	O	O	O
E13	andere Familienmitglieder zur Mithilfe im Haushalt anhalten	O	O	O	O	O	O
E14	mich zurückziehen	O	O	O	O	O	O
E15	mit dem Arzt oder der Ärztin über meine Sorgen um mein krankes Kind sprechen	O	O	O	O	O	O
E16	darauf vertrauen, dass die Klinik (das Behandlungszentrum) im besten Interesse meiner Familie handelt	O	O	O	O	O	O

	Wie hilfreich war oder ist für Sie …	gar nicht hilfreich	kaum hilfreich	etwas hilfreich	sehr hilfreich	habe ich nicht ausprobiert	war nicht möglich
E17	enge Beziehungen zu anderen Menschen aufbauen	O	O	O	O	O	O
E18	an Gott glauben	O	O	O	O	O	O
E19	mich selbst als Person weiterentwickeln	O	O	O	O	O	O
E20	mit anderen Eltern in vergleichbarer Situation sprechen und von ihren Erfahrungen lernen	O	O	O	O	O	O
E21	in der Familie mit allen etwas gemeinsam machen	O	O	O	O	O	O
E22	Zeit und Kraft in den Beruf einbringen	O	O	O	O	O	O
E23	daran glauben, dass mein Kind die bestmögliche medizinische Behandlung erhält	O	O	O	O	O	O
E24	Freunde nach Hause einladen	O	O	O	O	O	O
E25	darüber lesen, wie andere Menschen in meiner Situation damit umgehen	O	O	O	O	O	O
E26	mit Verwandten etwas unternehmen	O	O	O	O	O	O
E27	mehr Selbstvertrauen und Unabhängigkeit entwickeln	O	O	O	O	O	O
E28	daran denken, dass ich vieles habe, wofür ich dankbar sein kann	O	O	O	O	O	O
E29	Hobbies (Kunst, Musik, Sport usw.) betreiben	O	O	O	O	O	O
E30	Freunden und Nachbarn unsere Familiensituation erläutern, damit sie uns verstehen	O	O	O	O	O	O
E31	unser krankes Kind ermutigen, unabhängiger zu werden	O	O	O	O	O	O
E32	auf mein Äußeres achten, gepflegt bleiben	O	O	O	O	O	O
E33	mit Freunden regelmäßig etwas unternehmen (Partys usw.)	O	O	O	O	O	O
E34	regelmäßig mit meinem Partner / meiner Partnerin ausgehen	O	O	O	O	O	O
E35	sicherstellen, dass die verordnete Behandlung (Medikamente usw.) zu Hause täglich eingehalten wird	O	O	O	O	O	O
E36	die Beziehung zu meinem Partner / meiner Partnerin enger gestalten	O	O	O	O	O	O

	Wie hilfreich war oder ist für Sie …	gar nicht hilfreich	kaum hilfreich	etwas hilfreich	sehr hilfreich	habe ich nicht ausprobiert	war nicht möglich
E37	mir selbst erlauben, auch einmal ärgerlich zu sein	O	O	O	O	O	O
E38	mich verstärkt meinem Kind / meinen Kindern widmen	O	O	O	O	O	O
E39	mit jemandem (nicht Arzt/Ärztin oder Psychologe/Psychologin) über meine Gefühle sprechen	O	O	O	O	O	O
E40	mehr über die Krankheit meines Kindes lesen	O	O	O	O	O	O
E41	über eigene Gefühle und Ängste mit meinem Partner / meiner Partnerin reden	O	O	O	O	O	O
E42	freimachen von häuslichen Verpflichtungen, um mich zu erholen	O	O	O	O	O	O
E43	mein krankes Kind regelmäßig in der Klinik (im Behandlungszentrum) vorstellen	O	O	O	O	O	O
E44	darauf vertrauen, dass immer alles gutgeht	O	O	O	O	O	O
E45	etwas mit meinem Kind / meinen Kindern unternehmen	O	O	O	O	O	O

Über Ihre Bedürfnisse

Häufig wünschen sich Eltern von Kindern mit chronischen seltenen Erkrankungen mehr Unterstützung, Informationen oder Austausch- und Gesprächsmöglichkeiten. Bitte schätzen Sie bei den folgenden Fragen ein, wie stark die genannten Bedürfnisse bei Ihnen ausgeprägt sind.

	Ich wünsche mir …	keine Angabe	**gar nicht**	**wenig**	**mittelmäßig**	**ziemlich**	**sehr stark**
F1	… mehr Informationen über mögliche Diagnostikmethoden der Erkrankung meines Kindes zu bekommen	○	○	○	○	○	○
F2	… mehr Informationen über mögliche Therapien der Erkrankung meines Kindes zu bekommen	○	○	○	○	○	○
F3	… mehr Informationen über den Umgang mit Behörden (Ämter, Versicherungen) zu bekommen	○	○	○	○	○	○
F4	… mehr Informationen über den Umgang mit meinem erkrankten Kind zu bekommen	○	○	○	○	○	○
F5	… mich mit anderen betroffenen Eltern treffen zu können	○	○	○	○	○	○
F6	… mit meinem Partner / meiner Partnerin häufiger über meine Probleme reden zu können	○	○	○	○	○	○
F7	... mit Familienangehörigen häufiger über meine Probleme reden zu können	○	○	○	○	○	○
F8	… mit Freunden häufiger über meine Probleme reden zu können	○	○	○	○	○	○
F9	… mit anderen betroffenen Eltern über meine Probleme reden zu können	○	○	○	○	○	○
F10	… einen Ansprechpartner oder eine Ansprechpartnerin im Behandlungszentrum zu haben, mit dem/der ich über meine Probleme reden kann (Psychologe/Psychologin, Sozialpädagoge/Sozialpädagogin)	○	○	○	○	○	○
F11	... im Alltag mehr Entlastung durch meinen Partner / meine Partnerin zu bekommen	○	○	○	○	○	○
F12	… im Alltag mehr Entlastung durch Familienangehörige zu bekommen	○	○	○	○	○	○
F13	… im Alltag mehr Entlastung durch Freunde zu bekommen	○	○	○	○	○	○
F14	… mehr Zeit für mich selbst zu haben	○	○	○	○	○	○
F15	… mehr Zeit für meinen Partner / meine Partnerin zu haben	○	○	○	○	○	○
F16	… mehr Zeit für andere Familienangehörige zu haben	○	○	○	○	○	○
F17	… mehr Zeit für meine Freunde zu haben	○	○	○	○	○	○

**Bitte beantworten Sie die folgenden Fragen nur,
wenn ihre erkrankten Kinder weitere Geschwister haben.**

	Ich wünsche mir…	keine Angaben	**gar nicht**	**wenig**	**mittelmäßig**	**ziemlich**	**sehr stark**
F18	... mehr Informationen über den Umgang mit meinem anderen Kind / meinen anderen Kindern zu bekommen	○	○	○	○	○	○
F19	… mehr Zeit für mein anderes Kind / meine anderen Kinder zu haben	○	○	○	○	○	○

Möchten Sie etwas ergänzen oder anmerken?

Haben wir ggf. wichtige Themen vergessen?

VIELEN DANK!!!

11.2 Curriculum Vitae

-- Lebenslauf wurde aus datenschutzrechtlichen Gründen entfernt --

11.3 Affirmation in Lieu of an Oath

Ich versichere ausdrücklich, dass ich die Arbeit selbständig und ohne fremde Hilfe verfasst, andere als die von mir angegebenen Quellen und Hilfsmittel nicht benutzt und die aus den benutzten Werken wörtlich oder inhaltlich entnommenen Stellen einzeln nach Ausgabe (Auflage und Jahr des Erscheinens), Band und Seite des benutzten Werkes kenntlich gemacht habe.
Ferner versichere ich, dass ich die Dissertation bisher nicht einem Fachvertreter an einer anderen Hochschule zur Überprüfung vorgelegt oder mich anderweitig um Zulassung zur Promotion beworben habe.
Ich erkläre mich einverstanden, dass meine Dissertation vom Dekanat der Medizinischen Fakultät mit einer gängigen Software zur Erkennung von Plagiaten überprüft werden kann.

Unterschrift: ……